Polyalanine expansion and frameshift mutations of the in congenital central hypoventilation syndrome

Nature Genetics 33, 459-461 DOI: 10.1038/ng1130

Citation Report

#	Article	IF	CITATIONS
1	PHOX2A and PHOX2B genes are highly co-expressed in human neuroblastoma. International Journal of Oncology, 1992, 33, 985.	1.4	8
2	Molecular analysis of congenital central hypoventilation syndrome. Human Genetics, 2003, 114, 22-26.	1.8	174
3	Mechanics and Control of Ventilation. Surgery, 2003, 21, iii-vi.	0.1	2
4	Idiopathic congenital central hypoventilation syndrome: Analysis of genes pertinent to early autonomic nervous system embryologic development and identification of mutations in PHOX2b. American Journal of Medical Genetics Part A, 2003, 123A, 267-278.	2.4	335
5	Noninvasive ventilatory strategies in the management of a newborn infant and three children with congenital central hypoventilation syndrome. Pediatric Pulmonology, 2003, 36, 544-548.	1.0	74
6	A clinician's plea. Nature Genetics, 2003, 33, 440-442.	9.4	38
7	MafB deficiency causes defective respiratory rhythmogenesis and fatal central apnea at birth. Nature Neuroscience, 2003, 6, 1091-1100.	7.1	154
8	Genes modulating chemical breathing control: lessons from mutant animals. Respiratory Physiology and Neurobiology, 2003, 136, 105-114.	0.7	16
9	Phox2b controls the development of peripheral chemoreceptors and afferent visceral pathways. Development (Cambridge), 2003, 130, 6635-6642.	1.2	279
10	Noradrenergic neuronal development is impaired by mutation of the proneural HASH-1 gene in congenital central hypoventilation syndrome (Ondine's curse). Human Molecular Genetics, 2003, 12, 3173-3180.	1.4	72
11	Polymorphism, shared functions and convergent evolution of genes with sequences coding for polyalanine domains. Human Molecular Genetics, 2003, 12, 2967-2979.	1.4	103
13	Dynamic Expression of <i>RGS4</i> in the Developing Nervous System and Regulation by the Neural Type-Specific Transcription Factor Phox2b. Journal of Neuroscience, 2003, 23, 10613-10621.	1.7	50
14	fMRI signal changes in response to forced expiratory loading in congenital central hypoventilation syndrome. Journal of Applied Physiology, 2004, 97, 1897-1907.	1.2	59
15	Genetics of Congenital Central Hypoventilation Syndrome. American Journal of Respiratory and Critical Care Medicine, 2004, 170, 16-21.	2.5	76
16	Sudden Infant Death Syndrome: Case-Control Frequency Differences at Genes Pertinent to Early Autonomic Nervous System Embryologic Development. Pediatric Research, 2004, 56, 391-395.	1.1	99
17	Genetics and Early Disturbances of Breathing Control: The Genetics of Childhood Disease and Development: A Series of Review Articles. Pediatric Research, 2004, 55, 729-733.	1.1	64
18	Mouse Brain Organization Revealed Through Direct Genome-Scale TF Expression Analysis. Science, 2004, 306, 2255-2257.	6.0	390
19	A molecular pathogenesis for transcription factor associated poly-alanine tract expansions. Human Molecular Genetics, 2004, 13, 2351-2359.	1.4	139

#	Article	IF	CITATIONS
20	Association between schizophrenia with ocular misalignment and polyalanine length variation in PMX2B. Human Molecular Genetics, 2004, 13, 551-561.	1.4	64
21	PHOX2B mutations and polyalanine expansions correlate with the severity of the respiratory phenotype and associated symptoms in both congenital and late onset Central Hypoventilation syndrome. Journal of Medical Genetics, 2004, 41, 373-380.	1.5	248
22	Haploinsufficiency for Phox2b in mice causes dilated pupils and atrophy of the ciliary ganglion: mechanistic insights into human congenital central hypoventilation syndrome. Human Molecular Genetics, 2004, 13, 1433-1439.	1.4	31
23	Contribution of Hox genes to the diversity of the hindbrain sensory system. Development (Cambridge), 2004, 131, 1259-1266.	1.2	50
24	Polyalanine expansions in human. Human Molecular Genetics, 2004, 13, R235-R243.	1.4	131
25	Highly Recurrent RET Mutations and Novel Mutations in Genes of the Receptor Tyrosine Kinase and Endothelin Receptor B Pathways in Chinese Patients with Sporadic Hirschsprung Disease. Clinical Chemistry, 2004, 50, 93-100.	1.5	55
26	Mechanisms of non-Mendelian inheritance in genetic disease. Human Molecular Genetics, 2004, 13, R225-R233.	1.4	69
27	Studying the genetics of Hirschsprung's disease: unraveling an oligogenic disorder. Clinical Genetics, 2004, 67, 6-14.	1.0	93
28	Localizing a putative mutation as the major contributor to the development of sporadic Hirschsprung disease to the RET genomic sequence between the promoter region and exon 2. European Journal of Human Genetics, 2004, 12, 604-612.	1.4	47
29	Screening of the ARX gene in 682 retarded males. European Journal of Human Genetics, 2004, 12, 701-705.	1.4	52
30	The Phox2B homeobox gene is mutated in sporadic neuroblastomas. Oncogene, 2004, 23, 9280-9288.	2.6	112
31	Alanine tracts: the expanding story of human illness and trinucleotide repeats. Trends in Genetics, 2004, 20, 51-58.	2.9	202
32	Characterization of Xenopus Phox2a and Phox2b defines expression domains within the embryonic nervous system and early heart field. Gene Expression Patterns, 2004, 4, 601-607.	0.3	13
33	Developmental gene control of brainstem function: views from the embryo. Progress in Biophysics and Molecular Biology, 2004, 84, 89-106.	1.4	39
34	Cytokines in sudden infant death syndrome. Lancet Neurology, The, 2004, 3, 81.	4.9	3
35	PHOX2B gene mutation in a patient with late-onset central hypoventilation. Pediatric Pulmonology, 2004, 38, 349-351.	1.0	50
36	The investigation and care of children with congenital central hypoventilation syndrome. Current Paediatrics, 2004, 14, 354-360.	0.2	4
37	Genes and genetics in respiratory control. Paediatric Respiratory Reviews, 2004, 5, 166-172.	1.2	12

		Citation Ri	EPORT	
#	Article		IF	CITATIONS
38	Molecular genetics of Hirschsprung's disease. Seminars in Pediatric Surgery, 2004	, 13, 236-248.	0.5	30
39	Germline Mutations of the Paired–Like Homeobox 2B (PHOX2B) Gene in Neuroblast Journal of Human Genetics, 2004, 74, 761-764.	oma. American	2.6	288
40	Germline PHOX2B Mutation in Hereditary Neuroblastoma. American Journal of Human 75, 727-730.	Genetics, 2004,	2.6	233
41	Sudden Infant Death Syndrome Is Not Associated with the Mutation of PHOX2B Gene Causative Gene of Congenital Central Hypoventilation Syndrome. Tohoku Journal of Ex Medicine, 2004, 203, 65-68.	, a Major kperimental	0.5	37
42	The French Congenital Central Hypoventilation Syndrome Registry. Chest, 2005, 127,	72-79.	0.4	199
43	Autonomic Function in Children With Congenital Central Hypoventilation Syndrome a Families. Chest, 2005, 128, 2478-2484.	nd Their	0.4	42
45	An In Vitro Approach to Test the Possible Role of Candidate Factors in the Transcriptio of the <i>RET</i> Proto-Oncogene. Gene Expression, 2005, 12, 137-149.	nal Regulation	0.5	24
46	Retracing "Ondine's Curse― Neurosurgery, 2005, 57, 354-363.		0.6	34
47	Chapter 16 Central sleep apnea. Handbook of Clinical Neurophysiology, 2005, , 207-2	23.	0.0	0
48	Central Sleep Apnea. , 2005, , 249-262.			0
49	New evidence of baroreflex dysfunction in congenital central hypoventilation syndrom Science, 2005, 108, 215-216.	e. Clinical	1.8	3
50	Childhood solid tumours: a developmental disorder. Nature Reviews Cancer, 2005, 5, 4	481-488.	12.8	124
51	PHOX2B mutations and genetic predisposition to neuroblastoma. Oncogene, 2005, 2-	4, 3050-3053.	2.6	45
52	Molecular responses to acidosis of central chemosensitive neurons in brain. Cellular Si 2005, 17, 799-808.	gnalling,	1.7	17
53	Neuroanatomic deficits in congenital central hypoventilation syndrome. Journal of Cor Neurology, 2005, 487, 361-371.	nparative	0.9	83
54	A novel 17 bp deletion in thePHOX2B gene causes congenital central hypoventilation total aganglionosis of the small and large intestine. American Journal of Medical Genet 2005, 139A, 50-51.	syndrome with iics, Part A,	0.7	11
55	Congenital Syndromes Affecting Respiratory Control during Sleep. , 2005, , 517-527.			2
56	Hypercapnic Exposure in Congenital Central Hypoventilation Syndrome Reveals CNS R Control Mechanisms. Journal of Neurophysiology, 2005, 93, 1647-1658.	espiratory	0.9	114

#	ARTICLE Hypoxia reveals posterior thalamic, cerebellar, midbrain, and limbic deficits in congenital central	IF 1.2	CITATIONS
59	Genome-wide linkage identifies novel modifier loci of aganglionosis in the Sox10Dom model of Hirschsprung disease. Human Molecular Genetics, 2005, 14, 1549-1558.	1.4	37
60	Database of mRNA gene expression profiles of multiple human organs. Genome Research, 2005, 15, 443-450.	2.4	110
61	A novel susceptibility locus for Hirschsprung's disease maps to 4q31.3-q32.3. Journal of Medical Genetics, 2005, 43, e35-e35.	1.5	21
62	Mutations of the RET gene in isolated and syndromic Hirschsprung's disease in human disclose major and modifier alleles at a single locus. Journal of Medical Genetics, 2005, 43, 419-423.	1.5	48
63	Molecular consequences of PHOX2B missense, frameshift and alanine expansion mutations leading to autonomic dysfunction. Human Molecular Genetics, 2005, 14, 3697-3708.	1.4	135
64	Distinct pathogenetic mechanisms for PHOX2B associated polyalanine expansions and frameshift mutations in congenital central hypoventilation syndrome. Human Molecular Genetics, 2005, 14, 1815-1824.	1.4	106
65	Ancestral RET haplotype associated with Hirschsprung's disease shows linkage disequilibrium breakpoint at -1249. Journal of Medical Genetics, 2005, 42, 322-327.	1.5	26
66	Pediatric Disorders with Autonomic Dysfunction: What Role for PHOX2B?. Pediatric Research, 2005, 58, 1-6.	1.1	67
67	Sleep-disordered Breathing in Newborn Mice Heterozygous for the Transcription Factor Phox2b. American Journal of Respiratory and Critical Care Medicine, 2005, 172, 238-243.	2.5	58
68	Functional Brain Deficits in Congenital Central Hypoventilation Syndrome: Commentary on the articles by Woo et al. on page 510 and Macey et al. on page 500 Pediatric Research, 2005, 57, 471-472.	1.1	5
69	Aberrant Neural Responses to Cold Pressor Challenges in Congenital Central Hypoventilation Syndrome. Pediatric Research, 2005, 57, 500-509.	1.1	38
70	Genes for normal sleep and sleep disorders. Annals of Medicine, 2005, 37, 580-589.	1.5	62
71	Congenital Central Hypoventilation Syndrome and Hirschsprung's Disease in an Extremely Preterm Infant. Pediatrics, 2005, 115, e737-e738.	1.0	27
72	PHOX2B Regulates Its Own Expression by a Transcriptional Auto-regulatory Mechanism. Journal of Biological Chemistry, 2005, 280, 37439-37448.	1.6	37
73	FMRI Responses to Hyperoxia in Congenital Central Hypoventilation Syndrome. Pediatric Research, 2005, 57, 510-518.	1.1	43
74	PHOX2B Genotype Allows for Prediction of Tumor Risk in Congenital Central Hypoventilation Syndrome. American Journal of Human Genetics, 2005, 76, 421-426.	2.6	222
75	La mort subite du nourrisson(MSN) Données récentes en physiologie. Médecine Du Sommeil, 2005, 2, 18-24.	0.3	1

ARTICLE IF CITATIONS # Sensitive Detection of Polyalanine Expansions in PHOX2B by Polymerase Chain Reaction Using 1.2 15 76 Bisulfite-Converted DNA. Journal of Molecular Diagnostics, 2005, 7, 638-640. Multiple Endocrine Neoplasia Type 2B and Hirschsprung's Disease. Clinical Gastroenterology and 2.4 Hepatology, 2005, 3, 423-431 Mutations of brainstem transcription factors and central respiratory disorders. Trends in Molecular 78 3.5 38 Medicine, 2005, 11, 23-30. Transgenic expression of an expanded (GCG)13 repeat PABPN1 leads to weakness and coordination defects in mice. Neurobiology of Disease, 2005, 18, 528-536. PABPN1 overexpression leads to upregulation of genes encoding nuclear proteins that are sequestered in oculopharyngeal muscular dystrophy nuclear inclusions. Neurobiology of Disease, 80 2.1 70 2005, 18, 551-567. Variations in aganglionic segment length of the enteric neural plexus in Mowat-Wilson syndrome. Journal of Pediatric Surgery, 2005, 40, 1411-1419. 0.8 The other trinucleotide repeat: polyalanine expansion disorders. Current Opinion in Genetics and 83 1.5 156 Development, 2005, 15, 285-293. Germline mutations of the paired-like homeobox 2B (PHOX2B) gene in neuroblastoma. Cancer Letters, 84 3.2 2005, 228, 51-58. 85 Phox2B mutations and the Delta–Notch pathway in neuroblastoma. Cancer Letters, 2005, 228, 59-63. 3.2 27 Neurotrophic Factor Expression in Three Infants With Ondine's Curse. Pediatric Neurology, 2005, 33, 1.0 331-336. Genetics of normal and pathological sleep in humans. Sleep Medicine Reviews, 2005, 9, 91-100. 87 3.8 127 Regulation of respiratory neuron development by neurotrophic and transcriptional signaling mechanisms. Respiratory Physiology and Neurobiology, 2005, 149, 99-109. In pursuit (and discovery) of a genetic basis for congenital central hypoventilation syndrome. 89 0.7 60 Respiratory Physiology and Neurobiology, 2005, 149, 73-82. Short-term blood pressure and heart rate variability in congenital central hypoventilation syndrome (Ondine's curse). Clinical Science, 2005, 108, 225-230. 1.8 A Perfect Storm. New England Journal of Medicine, 2005, 353, 1956-1961. 92 13.9 3 Molecular Basis of Hirschsprung's Disease and Other Congenital Enteric Neuropathies. Seminars in Colon and Rectal Surgery, 2006, 17, 20-28. Transcriptional Regulatory Elements in the Human Genome. Annual Review of Genomics and Human 95 2.5724 Genetics, 2006, 7, 29-59. A Cook's tour around Hirschsprung's disease. Current Paediatrics, 2006, 16, 182-191.

#	Article	IF	CITATIONS
98	Polyalanine and Polyglutamine Diseases: Possible Common Mechanisms?. , 2006, , 487-513.		2
99	Disorders of Breathing during Sleep. , 2006, , 1046-1070.		3
100	PHOX2B mutations in three Chinese patients with congenital central hypoventilation syndrome. Chinese Medical Journal, 2006, 119, 1749-1752.	0.9	10
101	SLEEP DISORDERS Hypoventilation. , 2006, , 85-91.		1
102	Central Sleep Apnea, Hypoventilation Syndromes and Periodic Breathing Disorders. , 2006, 35, 180-191.		2
103	Expanding the phenotypic spectrum of L1CAM-associated disease. Clinical Genetics, 2006, 69, 414-419.	1.0	31
104	Case report of Haddad syndrome in a newborn: congenital central hypoventilation syndrome and Hirschsprung's disease. Journal of Perinatology, 2006, 26, 259-260.	0.9	6
105	Acute role of the brain-derived neurotrophic factor (BDNF) on the respiratory neural network activity in mice in vitro. Journal of Physiology (Paris), 2006, 100, 290-296.	2.1	3
106	Elevated mean diffusivity in widespread brain regions in congenital central hypoventilation syndrome. Journal of Magnetic Resonance Imaging, 2006, 24, 1252-1258.	1.9	40
107	PHOX2B analysis in non-syndromic neuroblastoma cases shows novel mutations and genotype–phenotype associations. American Journal of Medical Genetics, Part A, 2006, 140A, 1297-1301.	0.7	44
108	Sudden infant death syndrome: Case-control frequency differences in paired like homeobox (PHOX)2B gene. American Journal of Medical Genetics, Part A, 2006, 140A, 1687-1691.	0.7	22
109	The TLX2 homeobox gene is a transcriptional target of PHOX2B in neural-crest-derived cells. Biochemical Journal, 2006, 395, 355-361.	1.7	41
110	Neurological features of congenital fibrosis of the extraocular muscles type 2 with mutations in PHOX2A. Brain, 2006, 129, 2363-2374.	3.7	59
111	Brainstem Anomalies in Two Patients Affected by Congenital Central Hypoventilation Syndrome. American Journal of Respiratory and Critical Care Medicine, 2006, 174, 706-709.	2.5	21
113	Molecular Bases of Human Neurocristopathies. , 2006, 589, 213-234.		79
114	Ventilatory response to hyperoxia in newborn mice heterozygous for the transcription factor Phox2b. American Journal of Physiology - Regulatory Integrative and Comparative Physiology, 2006, 290, R1691-R1696.	0.9	34
115	Characterization of Dermatoglyphics in PHOX2B-Confirmed Congenital Central Hypoventilation Syndrome. Pediatrics, 2006, 118, e408-e414.	1.0	7
116	PHOX2BMutation–confirmed Congenital Central Hypoventilation Syndrome. American Journal of Respiratory and Critical Care Medicine, 2006, 174, 923-927.	2.5	125

#	Article	IF	CITATIONS
117	Transgenic Models to Study Disorders of Respiratory Control in Newborn Mice. ILAR Journal, 2006, 47, 15-21.	1.8	16
118	Congenital Central Hypoventilation Syndrome. American Journal of Respiratory and Critical Care Medicine, 2006, 174, 1139-1144.	2.5	238
119	Expression of Phox2b by Brainstem Neurons Involved in Chemosensory Integration in the Adult Rat. Journal of Neuroscience, 2006, 26, 10305-10314.	1.7	311
121	A complex additive model of inheritance for Hirschsprung disease is supported by both RET mutations and predisposing RET haplotypes. Genetics in Medicine, 2006, 8, 704-710.	1.1	29
122	Facial Phenotype in Children and Young Adults with PHOX2B–Determined Congenital Central Hypoventilation Syndrome: Quantitative Pattern of Dysmorphology. Pediatric Research, 2006, 59, 39-45.	1.1	58
123	Pediatric Autonomic Disorders. Pediatrics, 2006, 118, 309-321.	1.0	133
124	Late-onset central hypoventilation syndrome: a family genetic study. European Respiratory Journal, 2006, 29, 312-316.	3.1	44
125	TASK Channels Determine pH Sensitivity in Select Respiratory Neurons But Do Not Contribute to Central Respiratory Chemosensitivity. Journal of Neuroscience, 2007, 27, 14049-14058.	1.7	167
126	Rapid-Onset Obesity With Hypothalamic Dysfunction, Hypoventilation, and Autonomic Dysregulation Presenting in Childhood. Pediatrics, 2007, 120, e179-e188.	1.0	175
127	Effects of temperature on ventilatory response to hypercapnia in newborn mice heterozygous for transcription factor Phox2b. American Journal of Physiology - Regulatory Integrative and Comparative Physiology, 2007, 293, R2027-R2035.	0.9	17
128	Detection and Significance of Serum Protein Marker of Hirschsprung Disease. Pediatrics, 2007, 120, e56-e60.	1.0	8
129	Genomics of Sleep-disordered Breathing. Proceedings of the American Thoracic Society, 2007, 4, 121-126.	3.5	25
130	Fog-induced Cough with Impaired Respiratory Sensation in Congenital Central Hypoventilation Syndrome. American Journal of Respiratory and Critical Care Medicine, 2007, 176, 825-832.	2.5	24
131	Differential aggregation and functional impairment induced by polyalanine expansions in FOXL2, a transcription factor involved in cranio-facial and ovarian development. Human Molecular Genetics, 2007, 17, 1010-1019.	1.4	62
132	Central hypoventilation with PHOX2B expansion mutation presenting in adulthood. Thorax, 2007, 62, 919-920.	2.7	28
133	Hirschsprung disease, associated syndromes and genetics: a review. Journal of Medical Genetics, 2007, 45, 1-14.	1.5	848
134	An ancestral variant of Secretogranin II confers regulation by PHOX2 transcription factors and association with hypertension. Human Molecular Genetics, 2007, 16, 1752-1764.	1.4	29
135	Effects of movement and work load in patients with congenital central hypoventilation syndrome. European Journal of Cardiovascular Prevention and Rehabilitation, 2007, 14, 294-298.	3.1	8

ARTICLE IF CITATIONS Sleep-Related Hypoventilation/Hypoxemic Syndromes. Chest, 2007, 131, 1936-1948. 0.4 74 136 Abnormal inspiratory depth in Phox2a haploinsufficient mice. Neuroscience, 2007, 145, 384-392. 137 1.1 Methylation-associated PHOX2B gene silencing is a rare event in human neuroblastoma. European 138 1.3 20 Journal of Cancer, 2007, 43, 2366-2372. Oculopharyngeal muscular dystrophy: Recent advances in the understanding of the molecular pathogenic mechanisms and treatment strategies. Biochimica Et Biophysica Acta - Molecular Basis of 1.8 146 Disease, 2007, 1772, 173-185. Geldanamycin promotes nuclear localisation and clearance of PHOX2B misfolded proteins containing 140 1.2 34 polyalanine expansions. International Journal of Biochemistry and Cell Biology, 2007, 39, 327-339. Concomitant existence of total bowel aganglionosis and congenital central hypoventilation 0.8 16 syndrome in a neonate with PHOX2B gene mutation. Journal of Pediatric Surgery, 2007, 42, e9-e11. 142 Neuroblastoma. Lancet, The, 2007, 369, 2106-2120. 6.3 1,856 Congenital Central Hypoventilation Syndrome with PHOX2B Gene Mutation in a Taiwanese Infant. 143 0.8 Journal of the Formosan Medical Association, 2007, 106, 69-73. 145 Sleep Breathing Disorders., 2007, , 43-65. 3 Sudden Infant Death Syndrome: Review of implicated genetic factors. American Journal of Medical 146 Genetics, Part A, 2007, 143A, 771-788. Epistatic interactions with a common hypomorphicRET allele in syndromic Hirschsprung disease. 147 1.1 75 Human Mutation, 2007, 28, 790-796. Unequal crossover recombination – population screening forPHOX2B gene polyalanine polymorphism 148 1.3 using CE. Electrophoresis, 2007, 28, 894-899. Central nervous system distribution of the transcription factor Phox2b in the adult rat. Journal of 149 0.9 124 Comparative Neurology, 2007, 503, 627-641. Transcriptional repression coordinates the temporal switch from motor to serotonergic 7.1 neurogenesis. Nature Neuroscience, 2007, 10, 1433-1439. Transcriptional regulation of TLX2 and impaired intestinal innervation: possible role of the PHOX2A 152 22 1.4 and PHOX2B genes. European Journal of Human Genetics, 2007, 15, 848-855. Abnormalities of the nucleus and nuclear inclusions in neurodegenerative disease: a work in 1.8 74 progress. Neuropathology and Applied Neurobiology, 2007, 33, 2-42. British Paediatric Neurology Association Annual Meeting 2007. Developmental Medicine and Child 154 1.1 104 Neurology, 2007, 49, 1-52. Genetics of sleep and sleep disorders. Cellular and Molecular Life Sciences, 2007, 64, 1216-1226. 2.4

#	Article	IF	CITATIONS
156	Vagal and sympathetic heart rate and blood pressure control in adult onset PHOX2B mutation–confirmed congenital central hypoventilation syndrome. Clinical Autonomic Research, 2007, 17, 177-185.	1.4	28
157	Polymorphic length of FOXE1 alanine stretch: evidence for genetic susceptibility to thyroid dysgenesis. Human Genetics, 2007, 122, 467-476.	1.8	61
158	De novo polyalanine expansion of PHOX2B in congenital central hypoventilation syndrome: unequal sister chromatid exchange during paternal gametogenesis. Journal of Human Genetics, 2007, 52, 921-925.	1.1	15
159	Symptomatology, pathophysiology, diagnostic work-up, and treatment of Hirschsprung disease in infancy and childhood. Current Gastroenterology Reports, 2007, 9, 245-253.	1.1	54
160	Congenital central hypoventilation syndrome. Indian Journal of Pediatrics, 2007, 74, 953-955.	0.3	6
161	Congenital central hypoventilation syndrome with hyperinsulinism in a preterm infant. Journal of Human Genetics, 2008, 53, 573-577.	1.1	37
162	Congenital central hypoventilation syndrome: <i>PHOX2B</i> genotype determines risk for sudden death. Pediatric Pulmonology, 2008, 43, 77-86.	1.0	105
163	A novel missense mutation in the <i>PHOX2B</i> gene is associated with late onset central hypoventilation syndrome. Pediatric Pulmonology, 2008, 43, 1036-1039.	1.0	13
164	Expression of polyalanine stretches induces mitochondrial dysfunction. Journal of Neuroscience Research, 2008, 86, 1529-1537.	1.3	21
165	Homozygous mutation of the PHOX2B gene in congenital central hypoventilation syndrome (Ondine's) Tj ETQq1	1 0 7843] 1.1	4 rgBT /Ove
166	Parental origin and somatic mosaicism of PHOX2B mutations in Congenital Central Hypoventilation Syndrome. Human Mutation, 2008, 29, 206-206.	1.1	52
167	Compound effect of <i>PHOX2B</i> and <i>RET</i> gene variants in congenital central hypoventilation syndrome combined with Hirschsprung disease. American Journal of Medical Genetics, Part A, 2008, 146A, 1486-1489.	0.7	23
168	Identification of ALK as a major familial neuroblastoma predisposition gene. Nature, 2008, 455, 930-935.	13.7	1,207
169	A ringleader identified. Nature, 2008, 455, 883-884.	13.7	12
170	Theories of almost everything. Nature, 2008, 455, 884-885.	13.7	17
171	Provalence and functional consequence of PHOX2R mutations in neuroblastoma Oncogene 2008, 27		
	469-476.	2.6	137
172	469-476. Retrotrapezoid nucleus and central chemoreception. Journal of Physiology, 2008, 586, 2043-2048.	2.6	137

#	Article	IF	CITATIONS
174	Are children just small adults? The differences between paediatric and adult sleep medicine. Internal Medicine Journal, 2008, 38, 719-731.	0.5	15
175	PABPN1 polyalanine tract deletion and long expansions modify its aggregation pattern and expression. Experimental Cell Research, 2008, 314, 1652-1666.	1.2	38
176	Phox2b expression in the aldosterone-sensitive HSD2 neurons of the NTS. Brain Research, 2008, 1226, 82-88.	1.1	13
177	The Retrotrapezoid Nucleus and Central Chemoreception. Tzu Chi Medical Journal, 2008, 20, 239-242.	0.4	1
178	Congenital central hypoventilation syndrome (CCHS) and sudden infant death syndrome (SIDS): Kindred disorders of autonomic regulation. Respiratory Physiology and Neurobiology, 2008, 164, 38-48.	0.7	51
179	Noradrenergic modulation of the respiratory neural network. Respiratory Physiology and Neurobiology, 2008, 164, 123-130.	0.7	46
180	PHOX2B mutations and ventilatory control. Respiratory Physiology and Neurobiology, 2008, 164, 49-54.	0.7	22
181	The chemical neuroanatomy of breathing. Respiratory Physiology and Neurobiology, 2008, 164, 3-11.	0.7	170
182	Identification of neurotransmitters and co-localization of transmitters in brainstem respiratory neurons. Respiratory Physiology and Neurobiology, 2008, 164, 18-27.	0.7	34
183	A 2-Year Old With No Ventilator Requirement but Who Cannot Be Extubated. Seminars in Pediatric Neurology, 2008, 15, 157-159.	1.0	3
185	The Retrotrapezoid Nucleus and Central Chemoreception. Advances in Experimental Medicine and Biology, 2008, 605, 327-332.	0.8	32
186	Central Alveolar Hypoventilation Syndromes. Sleep Medicine Clinics, 2008, 3, 601-615.	1.2	24
187	Integration in Respiratory Control. Advances in Experimental Medicine and Biology, 2008, , .	0.8	2
190	The mutations and potential targets of the forkhead transcription factor FOXL2. Molecular and Cellular Endocrinology, 2008, 282, 2-11.	1.6	68
191	Hirschsprung's disease, acrocallosal syndrome, and congenital hydrocephalus: report of 2 patients and literature review. Journal of Pediatric Surgery, 2008, 43, e13-e17.	0.8	23
192	Polymorphisms in the genes encoding the 4 RET ligands, GDNF, NTN, ARTN, PSPN, and susceptibility to Hirschsprung disease. Journal of Pediatric Surgery, 2008, 43, 2042-2047.	0.8	15
193	The expression of PHOX2A, PHOX2B and of their target gene dopamine-β-hydroxylase (DβH) is not modified by exposure to extremely-low-frequency electromagnetic field (ELF-EMF) in a human neuronal model. Toxicology in Vitro, 2008, 22, 1489-1495.	1.1	13
195	Developmental Anatomy and Physiology of the Respiratory System. , 2008, , 15-34.		5

#	Article	IF	CITATIONS
196	Sleep-Disordered Breathing. , 2008, , 943-954.		0
197	A human mutation in Phox2b causes lack of CO ₂ chemosensitivity, fatal central apnea, and specific loss of parafacial neurons. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 1067-1072.	3.3	271
198	Transcription factor control of central respiratory neuron development. , 2008, , 191-221.		3
199	Sleep and Breathing in Children. , 0, , .		1
200	Carotid body oxygen sensing. European Respiratory Journal, 2008, 32, 1386-1398.	3.1	113
201	CO ₂ -Sensitive Preinspiratory Neurons of the Parafacial Respiratory Group Express Phox2b in the Neonatal Rat. Journal of Neuroscience, 2008, 28, 12845-12850.	1.7	141
202	Control of Precerebellar Neuron Development by <i>Olig3</i> bHLH Transcription Factor. Journal of Neuroscience, 2008, 28, 10124-10133.	1.7	54
203	<i>PHOX2B</i> Germline and Somatic Mutations in Late-Onset Central Hypoventilation Syndrome. American Journal of Respiratory and Critical Care Medicine, 2008, 177, 906-911.	2.5	85
204	Congenital Muscle Fiber-Type Disproportion in a Patient With Congenital Central Hypoventilation Syndrome Due to PHOX2B Mutations. Journal of Child Neurology, 2008, 23, 829-831.	0.7	5
205	Metrics of sequence constraint overlook regulatory sequences in an exhaustive analysis at <i>phox2b</i> . Genome Research, 2008, 18, 252-260.	2.4	101
206	Haddad Syndrome: A Case of an Infant With Central Congenital Hypoventilation Syndrome and Hirschsprung Disease. Journal of Child Neurology, 2008, 23, 341-343.	0.7	9
207	Neuropsychological, Behavioral, and Adaptive Functioning of Swiss Children With Congenital Central Hypoventilation Syndrome. Journal of Child Neurology, 2008, 23, 1254-1259.	0.7	22
208	Endocrine Manifestations of the Rapid-Onset Obesity with Hypoventilation, Hypothalamic, Autonomic Dysregulation, and Neural Tumor Syndrome in Childhood. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 3971-3980.	1.8	120
209	Development of the Neural Crest–Derived Intrinsic Innervation of the Human Lung. American Journal of Respiratory Cell and Molecular Biology, 2008, 38, 269-275.	1.4	20
210	Neural control of breathing: insights from genetic mouse models. Journal of Applied Physiology, 2008, 104, 1522-1530.	1.2	39
211	Effect of sleep stage on breathing in children with central hypoventilation. Journal of Applied Physiology, 2008, 105, 44-53.	1.2	55
212	Diffusion Tensor Imaging Demonstrates Brainstem and Cerebellar Abnormalities in Congenital Central Hypoventilation Syndrome. Pediatric Research, 2008, 64, 275-280.	1.1	87
213	Delineation of Late Onset Hypoventilation Associated with Hypothalamic Dysfunction Syndrome. Pediatric Research, 2008, 64, 689-694.	1.1	63

#	Article	IF	CITATIONS
214	The 2008 Carl Ludwig Lecture: retrotrapezoid nucleus, CO ₂ homeostasis, and breathing automaticity. Journal of Applied Physiology, 2008, 105, 404-416.	1.2	136
215	Mutation Analysis of Endothelinâ€B Receptor Gene in Patients With Hirschsprung Disease in Taiwan. Journal of Pediatric Gastroenterology and Nutrition, 2008, 46, 36-40.	0.9	7
216	Structural Abnormalities in the Brainstem and Cerebellum in Congenital Central Hypoventilation Syndrome: Commentary on the article by Kumar et al. on page 275. Pediatric Research, 2008, 64, 226-227.	1.1	4
217	Analysis of neuroblastoma tumour progression; loss of PHOX2B on 4p13 and 17q gain are early events in neuroblastoma tumourigenesis. International Journal of Oncology, 2008, , .	1.4	5
218	The Genetic Basis of Sleep Disorders. Current Pharmaceutical Design, 2008, 14, 3386-3395.	0.9	23
219	Rare Occurrence of PHOX2b Mutations in Sporadic Neuroblastomas. Journal of Pediatric Hematology/Oncology, 2008, 30, 728-732.	0.3	20
220	Transcription factors and the genetic organization of brain stem respiratory neurons. Journal of Applied Physiology, 2008, 104, 1513-1521.	1.2	86
221	Sleep, Breathing, and Neurologic Disorders. , 2009, , 436-498.		2
222	A developmental and genetic classification for midbrain-hindbrain malformations. Brain, 2009, 132, 3199-3230.	3.7	262
223	Defective Respiratory Rhythmogenesis and Loss of Central Chemosensitivity in Phox2b Mutants Targeting Retrotrapezoid Nucleus Neurons. Journal of Neuroscience, 2009, 29, 14836-14846.	1.7	115
224	DSCAM Deficiency Causes Loss of Pre-Inspiratory Neuron Synchroneity and Perinatal Death. Journal of Neuroscience, 2009, 29, 2984-2996.	1.7	36
225	Therapy of Hypoventilation. Seminars in Respiratory and Critical Care Medicine, 2009, 30, 359-366.	0.8	2
226	Central Congenital Hypoventilation Syndrome: Changing Face of a Less Mysterious but More Complex Genetic Disorder. Seminars in Respiratory and Critical Care Medicine, 2009, 30, 262-274.	0.8	13
227	Congenital Hypoventilation Syndromes. Seminars in Respiratory and Critical Care Medicine, 2009, 30, 339-347.	0.8	11
228	Mammillary Body and Fornix Injury in Congenital Central Hypoventilation Syndrome. Pediatric Research, 2009, 66, 429-434.	1.1	29
229	Breathing with <i>Phox2b</i> . Philosophical Transactions of the Royal Society B: Biological Sciences, 2009, 364, 2477-2483.	1.8	43
230	Retrotrapezoid nucleus, respiratory chemosensitivity and breathing automaticity. Respiratory Physiology and Neurobiology, 2009, 168, 59-68.	0.7	87
231	PHOX2B in respiratory control: Lessons from congenital central hypoventilation syndrome and its mouse models. Respiratory Physiology and Neurobiology, 2009, 168, 125-132.	0.7	63

#	Article	IF	CITATIONS
232	Phox2b, RTN/pFRG neurons and respiratory rhythmogenesis. Respiratory Physiology and Neurobiology, 2009, 168, 13-18.	0.7	74
233	Embryonic emergence of the respiratory rhythm generator. Respiratory Physiology and Neurobiology, 2009, 168, 86-91.	0.7	32
234	Asymmetrical distribution of non-conserved regulatory sequences at PHOX2B is reflected at the ENCODE loci and illuminates a possible genome-wide trend. BMC Genomics, 2009, 10, 8.	1.2	22
235	Sensing, physiological effects and molecular response to elevated CO ₂ levels in eukaryotes. Journal of Cellular and Molecular Medicine, 2009, 13, 4304-4318.	1.6	41
236	Autosomal recessive diseases among the Athabaskans of the Southwestern United States: Recent advances and implications for the future. American Journal of Medical Genetics, Part A, 2009, 149A, 2602-2611.	0.7	10
237	<i>In Vitro</i> studies of non poly alanine PHOX2B mutations argue against a loss-of-function mechanism for congenital central hypoventilation. Human Mutation, 2009, 30, E421-E431.	1.1	34
238	Interaction between PHOX2B and CREBBP mediates synergistic activation: Mechanistic implications of PHOX2B mutants. Human Mutation, 2009, 30, 655-660.	1.1	20
239	Galanin is a selective marker of the retrotrapezoid nucleus in rats. Journal of Comparative Neurology, 2009, 512, 373-383.	0.9	49
240	Genetic basis of Hirschsprung's disease. Pediatric Surgery International, 2009, 25, 543-558.	0.6	117
241	Cholinergic switch associated with morphological differentiation in neuroblastoma. Journal of Pathology, 2009, 219, 463-472.	2.1	26
242	Congenital central hypoventilation syndrome from past to future: Model for translational and transitional autonomic medicine. Pediatric Pulmonology, 2009, 44, 521-535.	1.0	99
243	Genetic identification of an embryonic parafacial oscillator coupling to the preBötzinger complex. Nature Neuroscience, 2009, 12, 1028-1035.	7.1	186
244	Math1 Is Essential for the Development of Hindbrain Neurons Critical for Perinatal Breathing. Neuron, 2009, 64, 341-354.	3.8	146
245	Transcriptional regulation of RET by Nkx2-1, Phox2b, Sox10, and Pax3. Journal of Pediatric Surgery, 2009, 44, 1904-1912.	0.8	54
246	Association analysis of the PHOX2B gene with Hirschsprung disease in the Han Chinese population of Southeastern China. Journal of Pediatric Surgery, 2009, 44, 1805-1811.	0.8	10
247	Dilated basilar arteries in patients with congenital central hypoventilation syndrome. Neuroscience Letters, 2009, 467, 139-143.	1.0	14
248	The emerging molecular pathogenesis of neuroblastoma: implications for improved risk assessment and targeted therapy. Genome Medicine, 2009, 1, 74.	3.6	34
249	PHOX2B Mutation-Confirmed Congenital Central Hypoventilation Syndrome in a Chinese Family. Chest, 2009, 135, 537-544.	0.4	36

#	Article	IF	CITATIONS
250	The Phox2 pathway is differentially expressed in neuroblastoma tumors, but no mutations were found in the candidate tumor suppressor gene PHOX2A. International Journal of Oncology, 2009, 34, 697-705.	1.4	12
251	Differences between three inbred rat strains in number of K+ channel-immunoreactive neurons in the medullary raphé nucleus. Journal of Applied Physiology, 2010, 108, 1003-1010.	1.2	5
252	Carbon dioxide chemoreception and hypoventilation syndromes with autonomic dysregulation. Journal of Applied Physiology, 2010, 108, 979-988.	1.2	35
253	Slowly progressive neuromuscular diseases. , 2010, , 362-372.		Ο
254	Novel neuropathologic findings in the Haddad syndrome. Acta Neuropathologica, 2010, 119, 261-269.	3.9	31
255	Fine mapping of the 9q31 Hirschsprung's disease locus. Human Genetics, 2010, 127, 675-683.	1.8	27
256	A new familial cancer syndrome including predisposition to Wilms tumor and neuroblastoma. Familial Cancer, 2010, 9, 425-430.	0.9	9
257	Development of ventilatory control in infants. Paediatric Respiratory Reviews, 2010, 11, 199-207.	1.2	64
258	Retrotrapezoid nucleus and parafacial respiratory group. Respiratory Physiology and Neurobiology, 2010, 173, 244-255.	0.7	85
259	Central chemoreception: Lessons from mouse and human genetics. Respiratory Physiology and Neurobiology, 2010, 173, 312-321.	0.7	28
260	Chemosensitivity recovery in Ondine's curse syndrome under treatment with desogestrel. Respiratory Physiology and Neurobiology, 2010, 171, 171-174.	0.7	47
261	lonic mechanisms of central CO2 chemosensitivity. Respiratory Physiology and Neurobiology, 2010, 173, 298-304.	0.7	3
262	Degeneracy as a substrate for respiratory regulation. Respiratory Physiology and Neurobiology, 2010, 172, 1-7.	0.7	23
263	Congenital central hypoventilation syndrome and the PHOX2B gene: A model of respiratory and autonomic dysregulation. Respiratory Physiology and Neurobiology, 2010, 173, 322-335.	0.7	65
264	From circuits to behaviour: motor networks in vertebrates. Current Opinion in Neurobiology, 2010, 20, 116-125.	2.0	64
265	Fos-Tau-LacZ mice reveal sex differences in brainstem c-fos activation in response to mild carbon dioxide exposure. Brain Research, 2010, 1311, 51-63.	1.1	10
266	Distribution and phenotype of Phox2aâ€containing neurons in the adult spragueâ€dawley rat. Journal of Comparative Neurology, 2010, 518, 2202-2220.	0.9	22
267	Central respiratory chemoreception. Journal of Comparative Neurology, 2010, 518, 3883-3906.	0.9	199

#	Article	IF	CITATIONS
268	MECP2 duplication in a patient with congenital central hypoventilation. American Journal of Medical Genetics, Part A, 2010, 152A, 1591-1593.	0.7	16
269	Novel MLPA procedure using self-designed probes allows comprehensive analysis for CNVs of the genes involved in Hirschsprung disease. BMC Medical Genetics, 2010, 11, 71.	2.1	6
270	Homologs of genes expressed in Caenorhabditis elegans GABAergic neurons are also found in the developing mouse forebrain. Neural Development, 2010, 5, 32.	1.1	4
271	Congenital central hypoventilation syndrome: Neurocognitive functioning in school age children. Pediatric Pulmonology, 2010, 45, 92-98.	1.0	56
272	Global genomic and RNA profiles for novel risk stratification of neuroblastoma. Cancer Science, 2010, 101, 2295-2301.	1.7	29
273	Molecular pathogenesis of peripheral neuroblastic tumors. Oncogene, 2010, 29, 1566-1579.	2.6	84
274	Congenital central hypoventilation syndrome: genotype–phenotype correlation in parents of affected children carrying a <i>PHOX2B</i> expansion mutation. Clinical Genetics, 2010, 78, 289-293.	1.0	19
275	Mutations in SCG10 Are Not Involved in Hirschsprung Disease. PLoS ONE, 2010, 5, e15144.	1.1	6
276	A Case of Congenital Central Hypoventilation Syndrome with PHOX2B Gene Mutation in a Korean Neonate. Journal of Korean Medical Science, 2010, 25, 1237.	1.1	4
277	Essential Role of Phox2b-Expressing Ventrolateral Brainstem Neurons in the Chemosensory Control of Inspiration and Expiration. Journal of Neuroscience, 2010, 30, 12466-12473.	1.7	136
278	Natural selection drives the accumulation of amino acid tandem repeats in human proteins. Genome Research, 2010, 20, 745-754.	2.4	88
279	Comparison of PHOX2B Testing Methods in the Diagnosis of Congenital Central Hypoventilation Syndrome and Mosaic Carriers. Diagnostic Molecular Pathology, 2010, 19, 224-231.	2.1	23
280	An Official ATS Clinical Policy Statement: Congenital Central Hypoventilation Syndrome. American Journal of Respiratory and Critical Care Medicine, 2010, 181, 626-644.	2.5	433
281	Neuroblastoma Phox2b Variants Stimulate Proliferation and Dedifferentiation of Immature Sympathetic Neurons. Journal of Neuroscience, 2010, 30, 905-915.	1.7	53
282	PHOX2B Immunolocalization of the Candidate Human Retrotrapezoid Nucleus. Pediatric and Developmental Pathology, 2010, 13, 291-299.	0.5	40
283	Task2 potassium channels set central respiratory CO ₂ and O ₂ sensitivity. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 2325-2330.	3.3	132
284	Polyalanine expansion of PHOX2B in congenital central hypoventilation syndrome: rs17884724:A>C is associated with 7-alanine expansion. Journal of Human Genetics, 2010, 55, 4-7.	1.1	7
285	Hypoventilation Syndromes. Clinics in Chest Medicine, 2010, 31, 249-270.	0.8	30

#	Article	IF	CITATIONS
286	Noninvasive Positive Pressure Ventilation in the Treatment of Hypoventilation in Children. Sleep Medicine Clinics, 2010, 5, 471-484.	1.2	6
287	Central Sleep Apnea. Clinics in Chest Medicine, 2010, 31, 235-248.	0.8	51
288	A Nonverbal Learning Disability in a Case of Central Hypoventilation Syndrome without aPHOX2BGene Mutation. Child Neuropsychology, 2010, 16, 202-208.	0.8	3
289	Contributions of pediatrics and pediatric pathology to the body of knowledge regarding human disease. Human Pathology, 2010, 41, 309-315.	1.1	3
290	Arousal response to hypoxia in newborns: Insights from animal models. Biological Psychology, 2010, 84, 39-45.	1.1	9
291	Central autonomic regulation in congenital central hypoventilation syndrome. Neuroscience, 2010, 167, 1249-1256.	1.1	38
292	Phox2b, congenital central hypoventilation syndrome and the control of respiration. Seminars in Cell and Developmental Biology, 2010, 21, 814-822.	2.3	37
293	Genetics and Genomics of Neuroblastoma. Cancer Treatment and Research, 2010, 155, 65-84.	0.2	84
294	Genetic factors determining the functional organization of neural circuits controlling rhythmic movements. Progress in Brain Research, 2010, 187, 39-46.	0.9	4
295	Postmortem molecular analysis to SIDS victims. Forensic Science International: Genetics Supplement Series, 2011, 3, e263-e264.	0.1	0
296	Oculopharyngeal muscular dystrophy. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2011, 101, 181-192.	1.0	26
297	Apnea of prematurity: pathogenesis and management strategies. Journal of Perinatology, 2011, 31, 302-310.	0.9	105
298	Advances in the understanding of constitutional and somatic genomic alterations in neuroblastoma. Cancer Genetics, 2011, 204, 113-121.	0.2	57
299	Home Mechanical Ventilation: A Canadian Thoracic Society Clinical Practice Guideline. Canadian Respiratory Journal, 2011, 18, 197-215.	0.8	182
300	Haddad Syndrome withPHOX2BGene Mutation in a Korean Infant. Journal of Korean Medical Science, 2011, 26, 312.	1.1	6
301	Executive Summary of Respiratory Indications for Polysomnography in Children: An Evidence-Based Review. Sleep, 2011, 34, 389-398.	0.6	152
302	Congenital Central Hypoventilation Syndrome. Advances in Neonatal Care, 2011, 11, 167-172.	0.5	0
303	Oncogenic mutations of <i>ALK</i> in neuroblastoma. Cancer Science, 2011, 102, 302-308.	1.7	49

#	Article	IF	CITATIONS
304	Contribution of excitatory amino acid receptors of the retrotrapezoid nucleus to the sympathetic chemoreflex in rats. Experimental Physiology, 2011, 96, 989-999.	0.9	33
305	The mechanics and control of ventilation. Surgery, 2011, 29, 212-216.	0.1	3
306	Prenatal development of central rhythm generation. Respiratory Physiology and Neurobiology, 2011, 178, 146-155.	0.7	31
307	Neuroblastoma aggressiveness in relation to sympathetic neuronal differentiation stage. Seminars in Cancer Biology, 2011, 21, 276-282.	4.3	28
308	Congenital Central Hypoventilation Syndrome in Children. Paediatric Respiratory Reviews, 2011, 12, 253-263.	1.2	50
309	Brainstem Tethering with Ondine's Curse. World Neurosurgery, 2011, 76, 592.e11-592.e14.	0.7	4
310	Congenital central hypoventilation syndrome due to PHOX2B mutation in a Saudi child: a case report. Sleep and Breathing, 2011, 15, 875-878.	0.9	3
311	PHOX2B mutations in patients with Ondine–Hirschsprung disease and a review of the literature. European Journal of Pediatrics, 2011, 170, 1267-1271.	1.3	18
312	Low amounts of PHOX2B expanded alleles in asymptomatic parents suggest unsuspected recurrence risk in congenital central hypoventilation syndrome. Journal of Molecular Medicine, 2011, 89, 505-513.	1.7	37
313	The paired-like homeobox 2B (PHOX2B) gene and respiratory control. Canadian Journal of Anaesthesia, 2011, 58, 1063-1068.	0.7	2
314	Descriptive analysis of central sleep apnea in childhood at a single center. Pediatric Pulmonology, 2011, 46, 1023-1030.	1.0	69
315	Homozygous <i>PTEN</i> deletion in neuroblastoma arising in a child with Cowden syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 1763-1766.	0.7	3
316	Rapid-Onset Obesity With Hypothalamic Dysfunction, Hypoventilation, and Autonomic Dysregulation: Analysis of Hypothalamic and Autonomic Candidate Genes. Pediatric Research, 2011, 70, 375-378.	1.1	66
317	Neuroblastoma Imaging. RoFo Fortschritte Auf Dem Gebiet Der Rontgenstrahlen Und Der Bildgebenden Verfahren, 2011, 183, 217-225.	0.7	12
318	Hirschsprung's disease in the neurologically challenged child. International Journal of Adolescent Medicine and Health, 2011, 23, 223-7.	0.6	4
319	Genetics of sleep disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2011, 99, 681-693.	1.0	7
320	Genetics of Sleep and Sleep Disorders in Humans. , 2011, , 184-198.		2
321	Central sleep apnea. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2011, 98, 411-419.	1.0	7

# 322	ARTICLE Decreased Cortical Thickness in Central Hypoventilation Syndrome. Cerebral Cortex, 2012, 22, 1728-1737.	IF 1.6	CITATIONS
323	ALK germline mutations in patients with neuroblastoma: a rare and weakly penetrant syndrome. European Journal of Human Genetics, 2012, 20, 291-297.	1.4	38
324	A reliable prognosis for congenital central hypoventilation syndrome should reflect a patient's genetic profile and management history. Pediatric Research, 2012, 72, 438-439.	1.1	1
325	Inheritance of polyalanine expansion mutation of PHOX2B in congenital central hypoventilation syndrome. Journal of Human Genetics, 2012, 57, 335-337.	1.1	23
326	Pontine Mechanisms of Respiratory Control. , 2012, 2, 2443-2469.		205
327	A Triple Threat: Down Syndrome, Congenital Central Hypoventilation Syndrome, and Hirschsprung Disease. Pediatrics, 2012, 130, e1382-e1384.	1.0	7
328	Response to Latorraca and Palli. Pediatric Research, 2012, 72, 439-440.	1.1	0
329	PHOX2B Immunolabeling. American Journal of Surgical Pathology, 2012, 36, 1141-1149.	2.1	55
330	Lower Brainstem Regulation of Visceral, Cardiovascular, and Respiratory Function. , 2012, , 1058-1073.		10
331	The E3 ubiquitin ligase TRIM11 mediates the degradation of congenital central hypoventilation syndrome-associated polyalanine-expanded PHOX2B. Journal of Molecular Medicine, 2012, 90, 1025-1035.	1.7	17
332	Chromosomal and related Mendelian Syndromes associated with Hirschsprung's disease. Pediatric Surgery International, 2012, 28, 1045-1058.	0.6	32
333	A case of congenital central hypoventilation syndrome. Journal of Anesthesia, 2012, 26, 922-924.	0.7	6
334	Late-onset, insidious course and invasive treatment of congenital central hypoventilation syndrome in a case with the Phox2B mutation: case report. Sleep and Breathing, 2012, 16, 951-955.	0.9	12
335	Developmental alterations of the respiratory human retrotrapezoid nucleus in sudden unexplained fetal and infant death. Autonomic Neuroscience: Basic and Clinical, 2012, 170, 12-19.	1.4	44
336	Genetic Diseases: Congenital Central Hypoventilation, Rett, and Praderâ€Willi Syndromes. , 2012, 2, 2, 2255-2279.		21
337	Genetics of human enteric neuropathies. Progress in Neurobiology, 2012, 96, 176-189.	2.8	36
338	Pupillometry in congenital central hypoventilation syndrome (CCHS): quantitative evidence of autonomic nervous system dysregulation. Pediatric Research, 2012, 71, 280-285.	1.1	41
339	Respiratory circuits: development, function and models. Current Opinion in Neurobiology, 2012, 22, 676-685.	2.0	30

#	Article	IF	CITATIONS
340	Congenital Central Hypoventilation Syndrome in a Full-Term Baby Presenting With Repeated Extubation Failure. Pediatrics and Neonatology, 2012, 53, 72-74.	0.3	3
341	Atoh1 Governs the Migration of Postmitotic Neurons that Shape Respiratory Effectiveness at Birth and Chemoresponsiveness in Adulthood. Neuron, 2012, 75, 799-809.	3.8	51
342	Adrenals. Medical Radiology, 2012, , 61-80.	0.0	2
343	Congenital Central Hypoventilation Syndrome with PHOX2B Gene Mutation. Indian Journal of Pediatrics, 2012, 79, 1526-1528.	0.3	5
347	A Single Gene Target of an ETS-Family Transcription Factor Determines Neuronal CO2-Chemosensitivity. PLoS ONE, 2012, 7, e34014.	1.1	38
348	Disorders of Breathing During Sleep. , 2012, , 1067-1086.		4
349	Genetic Factors Influencing the Risk and Clinical Outcome of Neuroblastoma. , 0, , .		0
350	Congenital Central Hypoventilation Syndrome (CCHS) and PHOX2B Mutations. , 2012, , 445-449.		2
351	Disorders of Central Respiratory Control During Sleep in Children. , 2012, , 434-447.		6
352	Variable human phenotype associated with novel deletions of the <i>PHOX2B</i> gene. Pediatric Pulmonology, 2012, 47, 153-161.	1.0	54
353	Germline mosaicism of <i>PHOX2B</i> mutation accounts for familial recurrence of congenital central hypoventilation syndrome (CCHS). American Journal of Medical Genetics, Part A, 2012, 158A, 2297-2301.	0.7	23
354	Advances in Molecular Genetics of Hirschsprung's Disease. Anatomical Record, 2012, 295, 1628-1638.	0.8	25
355	In vitro drug treatments reduce the deleterious effects of aggregates containing polyAla expanded PHOX2B proteins. Neurobiology of Disease, 2012, 45, 508-518.	2.1	32
356	Epidemiologic survey of patients with congenital central hypoventilation syndrome in Japan. Pediatrics International, 2012, 54, 123-126.	0.2	13
357	Hirschsprung disease: a developmental disorder of the enteric nervous system. Wiley Interdisciplinary Reviews: Developmental Biology, 2013, 2, 113-129.	5.9	120
358	Exome Sequencing Identified NRG3 as a Novel Susceptible Gene of Hirschsprung's Disease in a Chinese Population. Molecular Neurobiology, 2013, 47, 957-966.	1.9	30
359	Congenital Central Hypoventilation Syndrome with PHOX2B Gene Mutation: Are We Missing the Diagnosis?. Indian Journal of Pediatrics, 2013, 80, 688-690.	0.3	1
360	Obstructive sleep apnea syndrome (OSAS) in children with Class III malocclusion: involvement of the PHOX2B gene. Sleep and Breathing, 2013, 17, 1275-1280.	0.9	14

# 361	ARTICLE Gastrointestinal Tract and Hepatobiliary Duct System. , 2013, , 1-17.	IF	CITATIONS 0
362	Chemoreception and asphyxia-induced arousal. Respiratory Physiology and Neurobiology, 2013, 188, 333-343.	0.7	36
363	Contribution of rare and common variants determine complex diseases—Hirschsprung disease as a model. Developmental Biology, 2013, 382, 320-329.	0.9	119
365	Rodent models of sleep apnea. Respiratory Physiology and Neurobiology, 2013, 188, 355-361.	0.7	56
366	The respiratory chemoreception conundrum: Light at the end of the tunnel?. Brain Research, 2013, 1511, 126-137.	1.1	26
367	The developmental etiology and pathogenesis of Hirschsprung disease. Translational Research, 2013, 162, 1-15.	2.2	191
368	Autonomic dysfunction of glucose homoeostasis in congenital central hypoventilation syndrome. Acta Paediatrica, International Journal of Paediatrics, 2013, 102, e178-80.	0.7	15
369	Development and developmental disorders of the enteric nervous system. Nature Reviews Gastroenterology and Hepatology, 2013, 10, 43-57.	8.2	180
370	Intermittent Hyperglycemia due to Autonomic Nervous System Dysfunction: A New Feature in Patients with Congenital Central Hypoventilation Syndrome. Journal of Pediatrics, 2013, 162, 171-176.e2.	0.9	11
371	Anesthetic considerations for rapidâ€onset obesity, hypoventilation, hypothalamic dysfunction, and autonomic dysfunction (ROHHAD) syndrome in children. Paediatric Anaesthesia, 2013, 23, 28-32.	0.6	14
372	Congenital central hypoventilation syndrome. Respiratory Physiology and Neurobiology, 2013, 189, 272-279.	0.7	36
374	Congenital Central Hypoventilation Syndrome and Sudden Infant Death Syndrome: Disorders of Autonomic Regulation. Seminars in Pediatric Neurology, 2013, 20, 44-55.	1.0	25
375	Transcriptional dysregulation and impairment of PHOX2B auto-regulatory mechanism induced by polyalanine expansion mutations associated with congenital central hypoventilation syndrome. Neurobiology of Disease, 2013, 50, 187-200.	2.1	29
376	Building a brain in the gut: development ofÂthe enteric nervous system. Clinical Genetics, 2013, 83, 307-316.	1.0	141
377	Central Sleep Apnea. , 2013, 3, 141-163.		167
378	Peripheral–central chemoreceptor interaction and the significance of a critical period in the development of respiratory control. Respiratory Physiology and Neurobiology, 2013, 185, 156-169.	0.7	44
379	Peripheral chemoreceptors in congenital central hypoventilation syndrome. Respiratory Physiology and Neurobiology, 2013, 185, 186-193.	0.7	28
380	Distinct Neuroblastoma-associated Alterations of PHOX2B Impair Sympathetic Neuronal Differentiation in Zebrafish Models. PLoS Genetics, 2013, 9, e1003533.	1.5	55

	CITATION	Report	
#	Article	IF	CITATIONS
381	The unresolved puzzle why alanine extensions cause disease. Biological Chemistry, 2013, 394, 951-963.	1.2	9
382	Congenital Central Alveolar Hypoventilation Syndrome. , 2013, , 314-318.		0
383	A new device for the care of Congenital Central Hypoventilation Syndrome patients during sleep. , 2013, 2013, 2445-8.		2
384	Contributions of PHOX2B in the Pathogenesis of Hirschsprung Disease. PLoS ONE, 2013, 8, e54043.	1.1	30
385	Reversal of Pulmonary Hypertension after Diaphragm Pacing in an Adult Patient with Congenital Central Hypoventilation Syndrome. International Journal of Artificial Organs, 2013, 36, 434-438.	0.7	9
386	Diagnostic practices and disease surveillance in Canadian children with congenital central hypoventilation syndrome. Canadian Respiratory Journal, 2013, 20, 165-170.	0.8	4
387	Molecular oncology of neuroblastoma. , 0, , 669-678.		0
388	Congenital Central Hypoventilation Syndrome: A case report. Journal of Neonatology, 2013, 27, 39-40.	0.0	0
390	Structural and functional neuroimaging of congenital central hypoventilation syndrome. , 0, , 293-300.		0
391	Close Neighbours on Chromosome 2 the ALK and MYCN Genes. Implications for Targeted Therapeutics in Neuroblastoma. , 0, , .		0
392	Residual chemosensitivity to ventilatory challenges in genotyped congenital central hypoventilation syndrome. Journal of Applied Physiology, 2014, 116, 439-450.	1.2	39
393	A Case of Congenital Central Hypoventilation Syndrome with a Novel Mutation of the <i>PHOX2B</i> Gene Presenting as Central Sleep Apnea. Journal of Clinical Sleep Medicine, 2014, 10, 327-329.	1.4	15
396	Regulation of Breathing and Autonomic Outflows by Chemoreceptors. , 2014, 4, 1511-1562.		248
397	An Assistive Device for Congenital Central Hypoventilation Syndrome Outpatients During Sleep. Annals of Biomedical Engineering, 2014, 42, 2106-2116.	1.3	2
398	Genetic analysis of <i>PHOX2B</i> in sudden unexpected death in epilepsy cases. Neurology, 2014, 83, 1018-1021.	1.5	19
399	Mutations that disrupt PHOXB interaction with the neuronal calcium sensor HPCAL1 impede cellular differentiation in neuroblastoma. Oncogene, 2014, 33, 3316-3324.	2.6	25
400	Ophthalmologic Impairment during Adulthood in Central Congenital Hypoventilation Syndrome: A Longitudinal Cohort Analysis of Nine Patients. Ophthalmic Genetics, 2014, 35, 229-234.	0.5	6
401	Congenital Central Hypoventilation Syndrome. , 2014, , 301-305.		1

IF

ARTICLE

CITATIONS

402	Neurocristopathies. , 2014, , 361-394.		16
403	Developmental markers of ganglion cells in the enteric nervous system and their application for evaluation of <scp>H</scp> irschsprung disease. Pathology International, 2014, 64, 432-442.	0.6	8
404	Multiple congenital anomaliesâ€intellectual disability (MCAâ€iD) and neuroblastoma in a patient harboring a de novo 14q23.1q23.3 deletion. American Journal of Medical Genetics, Part A, 2014, 164, 1310-1317.	0.7	9
405	Congenital central hypoventilation syndrome and carbon dioxide sensitivity. European Journal of Pediatrics, 2014, 173, 1727-1730.	1.3	1
406	The progestin etonogestrel enhances the respiratory response to metabolic acidosis in newborn rats. Evidence for a mechanism involving supramedullary structures. Neuroscience Letters, 2014, 567, 63-67.	1.0	15
407	Central Hypoventilation Syndromes. Sleep Medicine Clinics, 2014, 9, 105-118.	1.2	38
408	PHOX2B polyalanine repeat length is associated with sudden infant death syndrome and unclassified sudden infant death in the Dutch population. International Journal of Legal Medicine, 2014, 128, 621-9.	1.2	20
409	Phox2bâ€expressing retrotrapezoid neurons and the integration of central and peripheral chemosensory control of breathing in conscious rats. Experimental Physiology, 2014, 99, 571-585.	0.9	70
410	Heterozygous 24-polyalanine repeats in the <i>PHOX2B</i> gene with different manifestations across three generations. Pediatric Pulmonology, 2014, 49, E13-E16.	1.0	15
411	Proneural bHLH Genes in Development and Disease. Current Topics in Developmental Biology, 2014, 110, 75-127.	1.0	65
412	Recent insights into the biology of neuroblastoma. International Journal of Cancer, 2014, 135, 2249-2261.	2.3	91
413	An unusual cause of fetal hypomobility:congenital central hypoventilation syndrome associated with hirschsprung disease. European Journal of Pediatrics, 2014, 173, 1607-1609.	1.3	3
414	Signaling molecules and transcription factors involved in the development of the sympathetic nervous system, with special emphasis on the superior cervical ganglion. Cell and Tissue Research, 2014, 357, 527-548.	1.5	25
415	Neural Crest Cells in Enteric Nervous System Development and Disease. , 2014, , 231-253.		0
416	Hypoventilation Syndromes of Infancy, Childhood, and Adulthood. Sleep Medicine Clinics, 2014, 9, 425-439.	1.2	2
417	Sleep Hypoventilation Syndromes and Noninvasive Ventilation in Children. Sleep Medicine Clinics, 2014, 9, 441-453.	1.2	0
418	Congenital hypoventilation syndrome and Hirschsprung's disease – Haddad syndrome: A neonatal case presentation. Journal of Neonatal-Perinatal Medicine, 2015, 8, 165-168.	0.4	4
419	Stateâ€dependent control of breathing by the retrotrapezoid nucleus. Journal of Physiology, 2015, 593, 2909-2926.	1.3	72

		15	C
#	ARTICLE	IF	CITATIONS
421	to PHOX2B mutations: a cross-sectional study. Respiratory Research, 2015, 16, 80.	1.4	14
422	Using non-invasive bi-level positive airway pressure ventilator via tracheostomy in children with congenital central hypoventilation syndrome: two case reports. Journal of Medical Case Reports, 2015, 9, 149.	0.4	4
423	Treatment of neuroblastoma in congenital central hypoventilation syndrome with a <i>PHOX2B</i> polyalanine repeat expansion mutation: New twist on a neurocristopathy syndrome. Pediatric Blood and Cancer, 2015, 62, 2007-2010.	0.8	12
424	Impaired neural structure and function contributing to autonomic symptoms in congenital central hypoventilation syndrome. Frontiers in Neuroscience, 2015, 9, 415.	1.4	32
425	A Phox2b BAC Transgenic Rat Line Useful for Understanding Respiratory Rhythm Generator Neural Circuitry. PLoS ONE, 2015, 10, e0132475.	1.1	23
426	Dysregulation of locus coeruleus development in congenital central hypoventilation syndrome. Acta Neuropathologica, 2015, 130, 171-183.	3.9	45
427	Diaphragm Pacing without Tracheostomy in Congenital Central Hypoventilation Syndrome Patients. Respiration, 2015, 89, 534-538.	1.2	41
428	Hyperplasia of pulmonary neuroendocrine cells in infancy and childhood. Seminars in Diagnostic Pathology, 2015, 32, 420-437.	1.0	42
429	Key Role of Amino Acid Repeat Expansions in the Functional Diversification of Duplicated Transcription Factors. Molecular Biology and Evolution, 2015, 32, 2263-2272.	3.5	24
430	Characterisation of novel RUNX2 mutation with alanine tract expansion from Japanese cleidocranial dysplasia patient. Mutagenesis, 2015, 31, gev057.	1.0	16
431	The RET Receptor Family. , 2015, , 559-591.		1
432	Thoracoscopic placement of phrenic nerve pacers for diaphragm pacing in congenital central hypoventilation syndrome. Journal of Pediatric Surgery, 2015, 50, 78-81.	0.8	18
433	Hypoxia Silences Retrotrapezoid Nucleus Respiratory Chemoreceptors via Alkalosis. Journal of Neuroscience, 2015, 35, 527-543.	1.7	60
434	Sleeping problems in mothers and fathers of patients suffering from congenital central hypoventilation syndrome. Sleep and Breathing, 2015, 19, 1057-1064.	0.9	11
435	A Newborn with Cyanosis and a Young Child with Hypoventilation. , 2015, , 427-430.		0
436	miR-204 mediates post-transcriptional down-regulation of PHOX2B gene expression in neuroblastoma cells. Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms, 2015, 1849, 1057-1065.	0.9	25
437	Identification of novel pathways and molecules able to down-regulate PHOX2B gene expression by in vitro drug screening approaches in neuroblastoma cells. Experimental Cell Research, 2015, 336, 43-57.	1.2	9
438	Abnormal auditory pathways in PHOX2B mutation positive congenital central hypoventilation syndrome. BMC Neurology, 2015, 15, 41.	0.8	6

#	ARTICLE	IF	CITATIONS
439	Endocrinology and Metabolism, 2015, 28, 705-8.	0.4	2
440	Dysgenesis of Enteroendocrine Cells in Aristalessâ€Related Homeobox Polyalanine Expansion Mutations. Journal of Pediatric Gastroenterology and Nutrition, 2015, 60, 192-199.	0.9	11
441	Nocturnal Non-Invasive Ventilation. , 2015, , .		2
442	Knockout mouse models of Hirschsprung's disease. Pediatric Surgery International, 2015, 31, 787-794.	0.6	15
443	Nocturnal Noninvasive Ventilation in Children. , 2015, , 135-161.		0
444	Genotype–phenotype relationship in Japanese patients with congenital central hypoventilation syndrome. Journal of Human Genetics, 2015, 60, 473-477.	1.1	43
445	That's not it, either-neither polymorphisms in PHOX2B nor in MIF are involved in sudden infant death syndrome (SIDS). International Journal of Legal Medicine, 2015, 129, 985-989.	1.2	10
446	Neural Control of Breathing and CO2 Homeostasis. Neuron, 2015, 87, 946-961.	3.8	340
447	Facing the challenge of mammalian neural microcircuits: taking a few breaths may help. Journal of Physiology, 2015, 593, 3-23.	1.3	87
448	Late-onset congenital central hypoventilation syndrome and a rare PHOX2B gene mutation. Sleep and Breathing, 2015, 19, 55-60.	0.9	20
449	Rapid-onset obesity, hypothalamic dysfunction, hypoventilation, and autonomic dysregulation in Saudi Arabia. Journal of King Abdulaziz University, Islamic Economics, 2016, 37, 1258-1260.	0.5	5
450	Chronic intestinal pseudoâ€obstruction in a child harboring a founder Hirschsprung RET mutation. American Journal of Medical Genetics, Part A, 2016, 170, 2400-2403.	0.7	5
451	Proton detection and breathing regulation by the retrotrapezoid nucleus. Journal of Physiology, 2016, 594, 1529-1551.	1.3	73
452	Congenital Central Hypoventilation Syndrome. Chest, 2016, 149, 809-815.	0.4	44
453	Preliminary Data on the Usability and Efficacy of an Assistive Device for the Congenital Central Hypoventilation Syndrome: An Observational Study. IFMBE Proceedings, 2016, , 451-456.	0.2	0
454	SÃndrome de hipoventilação central congênita associada à doença de Hirschsprung: relato de caso e revisão de literatura. Revista Paulista De Pediatria, 2016, 34, 374-378.	0.4	12
455	Clinical aspects of neurointestinal disease: Pathophysiology, diagnosis, and treatment. Developmental Biology, 2016, 417, 217-228.	0.9	65
456	Neuroblastoma and Its Zebrafish Model. Advances in Experimental Medicine and Biology, 2016, 916, 451-478.	0.8	16

#	Article	IF	CITATIONS
457	Respiratory and autonomic dysfunction in congenital central hypoventilation syndrome. Journal of Neurophysiology, 2016, 116, 742-752.	0.9	43
458	Role of Astrocytes in Central Respiratory Chemoreception. Advances in Experimental Medicine and Biology, 2016, 949, 109-145.	0.8	11
459	Glial Cells in Health and Disease of the CNS. Advances in Experimental Medicine and Biology, 2016, , .	0.8	9
460	Nalcn Is a "Leak" Sodium Channel That Regulates Excitability of Brainstem Chemosensory Neurons and Breathing. Journal of Neuroscience, 2016, 36, 8174-8187.	1.7	66
461	Advances in the translational genomics of neuroblastoma: From improving risk stratification and revealing novel biology to identifying actionable genomic alterations. Cancer, 2016, 122, 20-33.	2.0	175
462	Oncologic Phenotype of Peripheral Neuroblastic Tumors Associated With <i>PHOX2B</i> Nonâ€Polyalanine Repeat Expansion Mutations. Pediatric Blood and Cancer, 2016, 63, 71-77.	0.8	14
463	Mouse models of Hirschsprung disease and other developmental disorders of the enteric nervous system: Old and new players. Developmental Biology, 2016, 417, 139-157.	0.9	56
464	Caring for the Ventilator Dependent Child. Respiratory Medicine, 2016, , .	0.1	3
465	Neuroblastoma. Nature Reviews Disease Primers, 2016, 2, 16078.	18.1	907
466	Care of the Child with Congenital Central Hypoventilation Syndrome. Respiratory Medicine, 2016, , 331-353.	0.1	0
467	Area postrema undergoes dynamic postnatal changes in mice and humans. Journal of Comparative Neurology, 2016, 524, 1259-1269.	0.9	11
468	Could the retrotrapezoid nucleus neurons tell us something about SUDEP?. Epilepsy and Behavior, 2016, 61, 86-87.	0.9	0
469	Alanine Expansions Associated with Congenital Central Hypoventilation Syndrome Impair PHOX2B Homeodomain-mediated Dimerization and Nuclear Import. Journal of Biological Chemistry, 2016, 291, 13375-13393.	1.6	19
470	Congenital central hypoventilation syndrome associated with Hirschsprung's Disease: case report and literature review. Revista Paulista De Pediatria (English Edition), 2016, 34, 374-378.	0.3	9
471	Desogestrel enhances ventilation in ondine patients: Animal data involving serotoninergic systems. Neuropharmacology, 2016, 107, 339-350.	2.0	22
472	Pathophysiology of central sleep apneas. Sleep and Breathing, 2016, 20, 467-482.	0.9	31
473	Oxygen-sensing by arterial chemoreceptors: Mechanisms and medical translation. Molecular Aspects of Medicine, 2016, 47-48, 90-108.	2.7	50
474	SÃndrome ROHHAD (obesidad de rápida progresión, disfunción hipotalámica, hipoventilación y) Tj ETQq1 1 616-622.	0.784314 0.3	rgBT /Overlo 19

	Сітатіо	n Report	
#	Article	IF	CITATIONS
475	Neurological Complications of Respiratory Disease. Seminars in Pediatric Neurology, 2017, 24, 14-24.	1.0	3
476	Molecular Genetics of Neuroblastoma. , 2017, , 83-125.		Ο
477	Expanded polyalanine tracts function as nuclear export signals and promote protein mislocalization via eEF1A1 factor. Journal of Biological Chemistry, 2017, 292, 5784-5800.	1.6	18
478	Effects of RET, NRG1 and NRG3 Polymorphisms in a Chinese Population with Hirschsprung Disease. Scientific Reports, 2017, 7, 43222.	1.6	22
479	Intelligent volume-assured pressured support (iVAPS) for the treatment of congenital central hypoventilation syndrome. Sleep and Breathing, 2017, 21, 513-519.	0.9	27
480	Variable phenotype in a novel mutation in <i>PHOX2B</i> . American Journal of Medical Genetics, Part A, 2017, 173, 1705-1709.	0.7	16
481	Central Sleep Apnea, Hypoventilation Syndrome, and Sleep in High Altitude. , 2017, , 597-618.		0
482	Genetic susceptibility to neuroblastoma. Current Opinion in Genetics and Development, 2017, 42, 81-90.	1.5	75
483	Examination of <scp>PHOX</scp> 2B in adult neuroendocrine neoplasms reveals relatively frequent expression in phaeochromocytomas and paragangliomas. Histopathology, 2017, 71, 503-510.	1.6	13
484	Common PHOX2B poly-alanine contractions impair RET gene transcription, predisposing to Hirschsprung disease. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 1770-1777.	1.8	25
485	Brainstem mechanisms underlying the cough reflex and its regulation. Respiratory Physiology and Neurobiology, 2017, 243, 60-76.	0.7	34
486	Nonsense pathogenic variants in exon 1 of <i>PHOX2B</i> lead to translational reinitiation in congenital central hypoventilation syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 1200-1207.	0.7	24
487	Characterization of <i>Pabpn1</i> Expansion Mutations in a Large Cohort of Mexican Patients with Oculopharyngeal Muscular Dystrophy (Opmd). Journal of Investigative Medicine, 2017, 65, 705-708.	0.7	9
488	Embryonic hindbrain patterning genes delineate distinct cardio-respiratory and metabolic homeostatic populations in the adult. Scientific Reports, 2017, 7, 9117.	1.6	9
489	Case 2: Unexplained Hypercarbia in a Neonate in the Neonatal Intensive Care Unit. NeoReviews, 2017, 18, e668-e670.	0.4	0
490	Effects of NRG1 Polymorphisms on Hirschsprung's Disease Susceptibility: A Meta-analysis. Scientific Reports, 2017, 7, 9913.	1.6	7
491	Anesthetic Considerations for Patients With Congenital Central Hypoventilation Syndrome. Anesthesia and Analgesia, 2017, 124, 169-178.	1.1	16
492	Spanish patients with central hypoventilation syndrome included in the European Registry. The 2015 data. Anales De PediatrÃa (English Edition), 2017, 86, 255-263.	0.1	2

#	Article	IF	CITATIONS
493	Mutations in <i>MYO1H</i> cause a recessive form of central hypoventilation with autonomic dysfunction. Journal of Medical Genetics, 2017, 54, 754-761.	1.5	21
495	Phasic inhibition as a mechanism for generation of rapid respiratory rhythms. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, 12815-12820.	3.3	38
496	Rapid-onset obesity with hypothalamic dysregulation, hypoventilation, and autonomic dysregulation (ROHHAD syndrome): A case report and literature review. NeurologÃa (English Edition), 2017, 32, 616-622.	0.2	2
497	<scp>PHOX</scp> 2B reliably distinguishes neuroblastoma among small round blue cell tumours. Histopathology, 2017, 71, 786-794.	1.6	43
498	Distinctive features of Phox2b-expressing neurons in the rat reticular formation dorsal to the trigeminal motor nucleus. Neuroscience, 2017, 358, 211-226.	1.1	4
499	Defining the transcriptomic landscape of the developing enteric nervous system and its cellular environment. BMC Genomics, 2017, 18, 290.	1.2	38
500	Astrocyte-specific overexpressed gene signatures in response to methamphetamine exposure in vitro. Journal of Neuroinflammation, 2017, 14, 49.	3.1	34
501	Normal sleep on mechanical ventilation in adult patients with congenital central alveolar hypoventilation (Ondine's curse syndrome). Orphanet Journal of Rare Diseases, 2017, 12, 18.	1.2	8
502	The respiratory control mechanisms in the brainstem and spinal cord: integrative views of the neuroanatomy and neurophysiology. Journal of Physiological Sciences, 2017, 67, 45-62.	0.9	113
503	Congenital central hypoventilation syndrome: a bedside-to-bench success story for advancing early diagnosis and treatment and improved survival and quality of life. Pediatric Research, 2017, 81, 192-201.	1.1	51
505	Intrinsic Disorder in Proteins with Pathogenic Repeat Expansions. Molecules, 2017, 22, 2027.	1.7	49
506	Does the Polymorphism in the Length of the Polyalanine Tract of <i>FOXE1</i> Gene Influence the Risk of Thyroid Dysgenesis Occurrence?. Journal of Thyroid Research, 2017, 2017, 1-6.	0.5	5
507	Central Sleep Apnea. , 2017, , 1049-1058.e5.		1
508	Congenital central hypoventilation syndrome: diagnosis and management. Expert Review of Respiratory Medicine, 2018, 12, 283-292.	1.0	37
509	Analysis of sporadic neuroblastic tumors reveals a novel PHOX2B mutation in neuroblastoma. Gene Reports, 2018, 11, 239-243.	0.4	0
510	Breathing regulation and blood gas homeostasis after near complete lesions of the retrotrapezoid nucleus in adult rats. Journal of Physiology, 2018, 596, 2521-2545.	1.3	47
511	Clinical Presentation, Evaluation, and Management of Neuroblastoma. Pediatrics in Review, 2018, 39, 194-203.	0.2	17
512	Neuroblastoma. Japanese Journal of Clinical Oncology, 2018, 48, 214-241.	0.6	147

		CITATION REPO	RT	
#	Article	IF	-	CITATIONS
513	Genetic factors in sleep-disordered breathing. Respiratory Investigation, 2018, 56, 111-119.	0	.9	14
514	Congenital central hypoventilation syndrome: Broader cognitive deficits revealed by parent cont Pediatric Pulmonology, 2018, 53, 492-497.	rols. 1.	.0	16
515	The involvement of the pathway connecting the substantia nigra, the periaqueductal gray matter the retrotrapezoid nucleus in breathing control in a rat model of Parkinson's disease. Experiment Neurology, 2018, 302, 46-56.	r and tal 2	.0	36
516	Expanding the phenotype of congenital central hypoventilation syndrome impacts management decisions. American Journal of Medical Genetics, Part A, 2018, 176, 1398-1404.	O	.7	13
517	Congenital central hypoventilation syndrome mimicking mitochondrial disease. Clinical Case Rep (discontinued), 2018, 6, 465-468.	oorts 0	.2	2
518	Genetic susceptibility to neuroblastoma: current knowledge and future directions. Cell and Tissu Research, 2018, 372, 287-307.	e 1	.5	49
519	Genetic Background of Hirschsprung Disease: A Bridge Between Basic Science and Clinical Appli Journal of Cellular Biochemistry, 2018, 119, 28-33.	cation. 1.	.2	29
520	Neuropsychological profile and social cognition in congenital central hypoventilation syndrome (CCHS): Correlation with neuroimaging in a clinical case. Journal of Clinical and Experimental Neuropsychology, 2018, 40, 75-83.	0	.8	4
521	Congenital central hypoventilation syndrome: A pictorial demonstration of absent electrical diaphragmatic activity using nonâ€invasive neurally adjusted ventilatory assist. Journal of Paedia and Child Health, 2018, 54, 200-202.	trics 0	.4	5
522	Structural and functional differences in <i>PHOX2B</i> frameshift mutations underlie isolated or syndromic congenital central hypoventilation syndrome. Human Mutation, 2018, 39, 219-236.	1	.1	28
523	The genetics of congenital central hypoventilation syndrome: clinical implications. The Applicatio Clinical Genetics, 2018, Volume 11, 135-144.	on of 1.	.4	36
524	Advances in the molecular biology and pathogenesis of congenital central hypoventilation syndrome—implications for new therapeutic targets. Expert Opinion on Orphan Drugs, 2018, 6	5, 719-731. ^O	.5	6
525	Dysregulated glucose homeostasis in congenital central hypoventilation syndrome. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 1325-1333.	0	.4	5
526	Mutation in <i>LBX1/Lbx1</i> precludes transcription factor cooperativity and causes congenite hypoventilation in humans and mice. Proceedings of the National Academy of Sciences of the Un States of America, 2018, 115, 13021-13026.	l hited 3	.3	27
527	The advances of genetics research on Hirschsprung's disease. Pediatric Investigation, 2018, 2, 1	89-195. o	.6	4
528	Altered Expression of Phox2 Transcription Factors in the Locus Coeruleus in Major Depressive Disorder Mimicked by Chronic Stress and Corticosterone Treatment In Vivo and In Vitro. Neuroscience, 2018, 393, 123-137.	1.	.1	15
529	Systemic oxidative stress in congenital central hypoventilation syndrome. European Respiratory Journal, 2018, 52, 1801497.	3.	.1	6
530	Neurocristopathies: New insights 150 years after the neural crest discovery. Developmental Biol 2018, 444, S110-S143.	ogy, o	.9	136

#	Article	IF	CITATIONS
532	Desogestrel down-regulates PHOX2B and its target genes in progesterone responsive neuroblastoma cells. Experimental Cell Research, 2018, 370, 671-679.	1.2	12
533	Enteric nervous system development: what could possibly go wrong?. Nature Reviews Neuroscience, 2018, 19, 552-565.	4.9	128
534	Genetic Factors in Sleep Disorders: What Are the Roles of Genetic Factors in the Pathogenesis of Sleep Disorders?. Respiratory Disease Series, 2018, , 225-254.	0.1	1
535	Diagnosis, management and pathophysiology of central sleep apnea in children. Paediatric Respiratory Reviews, 2019, 30, 49-57.	1.2	41
536	Disorders of Breathing During Sleep. , 2019, , 1143-1159.e9.		1
537	The role of the hypothalamus in modulation of respiration. Respiratory Physiology and Neurobiology, 2019, 265, 172-179.	0.7	57
538	SÃndrome de Ondina (hipoventilación central congénita), una amenaza contra la vida al dormir. Iatreia, 2019, 32, 243-247.	0.1	0
539	Neonatal Congenital Central Hypoventilation Syndrome: Why We Should not Sleep on it. Literature Review of Forty-two Neonatal Onset Cases. Current Pediatric Reviews, 2019, 15, 139-153.	0.4	13
540	Short Tandem Repeat Expansions and RNA-Mediated Pathogenesis in Myotonic Dystrophy. International Journal of Molecular Sciences, 2019, 20, 3365.	1.8	69
541	Congenital malformation syndromes associated with peripheral neuroblastic tumors: A systematic review. Pediatric Blood and Cancer, 2019, 66, e27901.	0.8	5
542	The Retrotrapezoid Nucleus: Central Chemoreceptor and Regulator of Breathing Automaticity. Trends in Neurosciences, 2019, 42, 807-824.	4.2	129
543	Optogenetic analysis of respiratory neuronal networks in the ventral medulla of neonatal rats producing channelrhodopsin in Phox2b-positive cells. Pflugers Archiv European Journal of Physiology, 2019, 471, 1419-1439.	1.3	5
544	Congenital central hypoventilation syndrome: Severe disease caused by coâ€occurrence of two PHOX2B variants inherited separately from asymptomatic family members. American Journal of Medical Genetics, Part A, 2019, 179, 503-506.	0.7	10
545	The role of PHOX2Bâ€derived astrocytes in chemosensory control of breathing and sleep homeostasis. Journal of Physiology, 2019, 597, 2225-2251.	1.3	27
546	ExpansionHunter: a sequence-graph-based tool to analyze variation in short tandem repeat regions. Bioinformatics, 2019, 35, 4754-4756.	1.8	183
547	Animal Models of Aganglionosis. , 2019, , 97-113.		0
548	Functional Anatomy of theÂEnteric Nervous System. , 2019, , 31-76.		7
549	Congenital Anomalies and Genetic Associations in Hirschsprung's Disease. , 2019, , 175-199.		1

#	Article	IF	CITATIONS
550	Tridimensional mapping of Phox2b expressing neurons in the brainstem of adult <i>Macaca fascicularis</i> and identification of the retrotrapezoid nucleus. Journal of Comparative Neurology, 2019, 527, 2875-2884.	0.9	8
551	Two novel mutations in exon 3 of PHOX2B gene: think about congenital central hypoventilation syndrome in patients with Hirschsprung disease. Italian Journal of Pediatrics, 2019, 45, 49.	1.0	3
552	Chemosensitivity of <i>Phox2b</i> â€expressing retrotrapezoid neurons is mediated in part by input from 5â€HT neurons. Journal of Physiology, 2019, 597, 2741-2766.	1.3	38
553	Area Postrema: Fetal Maturation, Tumors, Vomiting Center, Growth, Role in Neuromyelitis Optica. Pediatric Neurology, 2019, 94, 21-31.	1.0	6
554	Congenital central hypoventilation syndrome and Hirschsprung disease: A retrospective review of the French National Registry Center on 33 cases. Journal of Pediatric Surgery, 2019, 54, 2325-2330.	0.8	12
555	The diverse neural crest: from embryology to human pathology. Development (Cambridge), 2019, 146, .	1.2	82
556	Massive Amniotic Fluid Aspiration in a Case of Sudden Neonatal Death With Severe Hypoplasia of the Retrotrapezoid/Parafacial Respiratory Group. Frontiers in Pediatrics, 2019, 7, 116.	0.9	5
557	NovelPHOX2Bmutations in congenital central hypoventilation syndrome. Pediatrics International, 2019, 61, 393-396.	0.2	7
558	A Novel c.676_677insG PHOX2B Mutation in Congenital Central Hypoventilation Syndrome. Journal of Clinical Sleep Medicine, 2019, 15, 509-513.	1.4	2
559	Breathing under Anesthesia. Anesthesiology, 2019, 130, 995-1006.	1.3	9
559 560	Breathing under Anesthesia. Anesthesiology, 2019, 130, 995-1006. Orexin Neurons Contribute to Central Modulation of Respiratory Drive by Progestins on ex vivo Newborn Rodent Preparations. Frontiers in Physiology, 2019, 10, 1200.	1.3 1.3	9
559 560 561	Breathing under Anesthesia. Anesthesiology, 2019, 130, 995-1006. Orexin Neurons Contribute to Central Modulation of Respiratory Drive by Progestins on ex vivo Newborn Rodent Preparations. Frontiers in Physiology, 2019, 10, 1200. A novel PHOX2B gene mutation in an extremely low birth weight infant with congenital central hypoventilation syndrome and variant Hirschsprung's disease. European Journal of Medical Genetics, 2019, 62, 103541.	1.3 1.3 0.7	9 13 5
559 560 561 563	Breathing under Anesthesia. Anesthesiology, 2019, 130, 995-1006. Orexin Neurons Contribute to Central Modulation of Respiratory Drive by Progestins on ex vivo Newborn Rodent Preparations. Frontiers in Physiology, 2019, 10, 1200. A novel PHOX2B gene mutation in an extremely low birth weight infant with congenital central hypoventilation syndrome and variant Hirschsprung's disease. European Journal of Medical Genetics, 2019, 62, 103541. Causative and common <i>PHOX2B Causative and common <i>PHOX2B</i></i>	1.3 1.3 0.7 1.0	9 13 5 39
559 560 561 563	Breathing under Anesthesia. Anesthesiology, 2019, 130, 995-1006. Orexin Neurons Contribute to Central Modulation of Respiratory Drive by Progestins on ex vivo Newborn Rodent Preparations. Frontiers in Physiology, 2019, 10, 1200. A novel PHOX2B gene mutation in an extremely low birth weight infant with congenital central hypoventilation syndrome and variant Hirschsprung's disease. European Journal of Medical Genetics, 2019, 62, 103541. Causative and common <i>PHOX2B Causative and common <i>PHOX2B PHOX2B Genetic predisposition and chromosome instability in neuroblastoma. Cancer and Metastasis Reviews, 2020, 39, 275-285.</i></i>	1.3 1.3 0.7 1.0 2.7	9 13 5 39 65
 559 560 561 563 564 565 	Breathing under Anesthesia. Anesthesiology, 2019, 130, 995-1006. Orexin Neurons Contribute to Central Modulation of Respiratory Drive by Progestins on ex vivo Newborn Rodent Preparations. Frontiers in Physiology, 2019, 10, 1200. A novel PHOX2B gene mutation in an extremely low birth weight infant with congenital central hypoventilation syndrome and variant Hirschsprung's disease. European Journal of Medical Genetics, 2019, 62, 103541. Causative and common <i>PHOX2B Genetic predisposition and chromosome instability in neuroblastoma. Cancer and Metastasis Reviews, 2020, 39, 275-285. Development of the brainstem respiratory circuit. Wiley Interdisciplinary Reviews: Developmental Biology, 2020, 9, e366.</i>	1.3 1.3 0.7 1.0 2.7 5.9	 9 13 5 39 65 16
 559 560 561 563 565 566 	Breathing under Anesthesia. Anesthesiology, 2019, 130, 995-1006. Orexin Neurons Contribute to Central Modulation of Respiratory Drive by Progestins on ex vivo Newborn Rodent Preparations. Frontiers in Physiology, 2019, 10, 1200. A novel PHOX2B gene mutation in an extremely low birth weight infant with congenital central hypoventilation syndrome and variant Hirschsprung's disease. European Journal of Medical Genetics, 2019, 62, 103541. Causative and common <i>PHOX2B PHOX2B Genetic predisposition and chromosome instability in neuroblastoma. Cancer and Metastasis Reviews, 2020, 39, 275-285. Development of the brainstem respiratory circuit. Wiley Interdisciplinary Reviews: Developmental Biology, 2020, 9, e366. Systematic Review of the Genetics of Sudden Unexpected Death in Epilepsy: Potential Overlap With Sudden Cardiac Death and Arrhythmiaa€Related Cenes. Journal of the American Heart Association, 2020, 9, e012264.</i>	1.3 1.3 0.7 1.0 2.7 5.9 1.6	9 13 5 39 65 16
 559 560 563 564 565 566 567 	Breathing under Anesthesia. Anesthesiology, 2019, 130, 995-1006. Orexin Neurons Contribute to Central Modulation of Respiratory Drive by Progestins on ex vivo Newborn Rodent Preparations. Frontiers in Physiology, 2019, 10, 1200. A novel PHOX2B gene mutation in an extremely low birth weight infant with congenital central hypoventilation syndrome and variant Hirschsprung's disease. European Journal of Medical Genetics, 2019, 62, 103541. Causative and common <i>PHOX2B Genetic predisposition and chromosome instability in neuroblastoma. Cancer and Metastasis Reviews, 2020, 39, 275-285. Development of the brainstem respiratory circuit. Wiley Interdisciplinary Reviews: Developmental Biology, 2020, 9, e366. Systematic Review of the Genetics of Sudden Unexpected Death in Epilepsy: Potential Overlap With Sudden Cardiac Death and ArrhythmiaäcRelated Genes. Journal of the American Heart Association, 2020, 9, e012264. Guidelines for diagnosis and management of congenital central hypoventilation syndrome. Orphanet Journal of Rare Diseases, 2020, 15, 252.</i>	1.3 1.3 0.7 1.0 2.7 5.9 1.6 1.2	 9 13 5 39 65 16 66 74

# 569	ARTICLE Neurodevelopmental outcome and respiratory management of congenital central hypoventilation syndrome: a retrospective study. BMC Pediatrics, 2020, 20, 342.	IF 0.7	CITATIONS
570	Neurocognition in Congenital Central Hypoventilation Syndrome: influence of genotype and ventilation method. Orphanet Journal of Rare Diseases, 2020, 15, 322.	1.2	11
571	A Rare Cause of Sleep-Disordered Breathing: ROHHAD Syndrome. Frontiers in Pediatrics, 2020, 8, 573227.	0.9	6
572	Vertebrate Sensory Ganglia: Common and Divergent Features of the Transcriptional Programs Generating Their Functional Specialization. Frontiers in Cell and Developmental Biology, 2020, 8, 587699.	1.8	45
573	Genetic Predisposition to Solid Pediatric Cancers. Frontiers in Oncology, 2020, 10, 590033.	1.3	31
574	Cough: Pathophysiology, Diagnosis and Treatment. , 2020, , .		4
575	Obesity Hypoventilation. Sleep Medicine Clinics, 2020, 15, 449-459.	1.2	2
576	Generation and characterization of iPSC lines (BGUi004-A, BGUi005-A) from two identical twins with polyalanine expansion in the paired-like homeobox 2B (PHOX2B) gene. Stem Cell Research, 2020, 48, 101955.	0.3	11
577	Sox10 Is a Specific Biomarker for Neural Crest Stem Cells in Immunohistochemical Staining in Wistar Rats. Disease Markers, 2020, 2020, 1-7.	0.6	3
578	Molecular Mechanisms of Acute Oxygen Sensing by Arterial Chemoreceptor Cells. Role of Hif2α. Frontiers in Physiology, 2020, 11, 614893.	1.3	6
579	Mechanisms of Genome Protection and Repair. Advances in Experimental Medicine and Biology, 2020, , .	0.8	2
580	Long Term Non-invasive Ventilation in Children With Central Hypoventilation. Frontiers in Pediatrics, 2020, 8, 288.	0.9	4
581	Adult-onset congenital central hypoventilation syndrome due to PHOX2B mutation. Acta Neurologica Belgica, 2021, 121, 23-35.	0.5	3
582	Neonatal apneic phenotype in a murine congenital central hypoventilation syndrome model is induced through nonâ€cell autonomous developmental mechanisms. Brain Pathology, 2021, 31, 84-102.	2.1	16
583	Ultrahigh-Frequency Echocardiography of Autonomic Devoid Phox2B Homozygous Embryos Does Not Reveal a Significant Cardiac Phenotype before Embryo Death. Ultrasound in Medicine and Biology, 2021, 47, 751-758.	0.7	1
584	Hirschsprung disease and other gastrointestinal motility disorders in patients with CCHS. European Journal of Pediatrics, 2021, 180, 469-473.	1.3	11
585	Research Advances on Therapeutic Approaches to Congenital Central Hypoventilation Syndrome (CCHS). Frontiers in Neuroscience, 2020, 14, 615666.	1.4	19
586	Ablation of Zfhx4 results in early postnatal lethality by disrupting the respiratory center in mice. Journal of Molecular Cell Biology, 2021, 13, 210-224.	1.5	2

#	ARTICLE	IF	CITATIONS
587	Sleep medicine and breathing control disorders. , 2021, , 694-738.		0
589	Congenital central hypoventilation syndrome and ventilatory responses during cardiopulmonary exercise testing. Pediatric Pulmonology, 2021, 56, 1694-1703.	1.0	4
590	How the Management of Children With Congenital Central Hypoventilation Syndrome Has Changed Over Time: Two Decades of Experience From an Italian Center. Frontiers in Pediatrics, 2021, 9, 648927.	0.9	9
591	A Common 3′UTR Variant of the PHOX2B Gene Is Associated With Infant Life-Threatening and Sudden Death Events in the Italian Population. Frontiers in Neurology, 2021, 12, 642735.	1.1	10
592	Mechanisms of repeat-associated non-AUG translation in neurological microsatellite expansion disorders. Biochemical Society Transactions, 2021, 49, 775-792.	1.6	12
593	Chemoreceptor mechanisms regulating CO ₂ â€induced arousal from sleep. Journal of Physiology, 2021, 599, 2559-2571.	1.3	6
594	Paired-like homeobox gene (PHOX2B) nonpolyalanine repeat expansion mutations (NPARMs): genotype–phenotype correlation in congenital central hypoventilation syndrome (CCHS). Genetics in Medicine, 2021, 23, 1656-1663.	1.1	16
595	Altered Mental Status and Respiratory Failure in an 11-Year-Old Female. Cureus, 2021, 13, e15164.	0.2	0
596	30 years of repeat expansion disorders: What have we learned and what are the remaining challenges?. American Journal of Human Genetics, 2021, 108, 764-785.	2.6	170
597	Parafacial neurons in the human brainstem express specific markers for neurons of the retrotrapezoid nucleus. Journal of Comparative Neurology, 2021, 529, 3313-3320.	0.9	4
599	An update on the neurological short tandem repeat expansion disorders and the emergence of long-read sequencing diagnostics. Acta Neuropathologica Communications, 2021, 9, 98.	2.4	83
600	Literature review: enteric nervous system development, genetic and epigenetic regulation in the etiology of Hirschsprung's disease. Heliyon, 2021, 7, e07308.	1.4	7
601	Genetic mutation in Hirschsprungs/congenital central hypoventilation syndrome. Journal of Pediatric Surgery Case Reports, 2021, 69, 101861.	0.1	1
602	Carotid body chemoreceptors: physiology, pathology, and implications for health and disease. Physiological Reviews, 2021, 101, 1177-1235.	13.1	85
604	Size matters: Large copy number losses in Hirschsprung disease patients reveal genes involved in enteric nervous system development. PLoS Genetics, 2021, 17, e1009698.	1.5	14
605	Obstructive Apneas in a Mouse Model of Congenital Central Hypoventilation Syndrome. American Journal of Respiratory and Critical Care Medicine, 2021, 204, 1200-1210.	2.5	11
606	Therapeutically targeting oncogenic CRCs facilitates induced differentiation of NB by RA and the BET bromodomain inhibitor. Molecular Therapy - Oncolytics, 2021, 23, 181-191.	2.0	6
607	Mutations of PHOX2B Gene in Patients of Obesity Hypoventilation Syndrome in Central India. Journal of Laboratory Physicians, 0, , .	0.4	0

		CITATION R	EPORT	
# 608	ARTICLE The Genetics of Sleep Disorders in Children: A Narrative Review. Brain Sciences, 2021,	11, 1259.	IF 1.1	CITATIONS
609	ACE2 expression in rat brain: Implications for COVID-19 associated neurological manif Experimental Neurology, 2021, 345, 113837.	estations.	2.0	50
610	Disorders of Respiratory Control and Central Hypoventilation Syndromes. , 2021, , 363	3-377.		0
611	Central sleep apnoeas and hypoventilation syndromes. , 2021, , 725-731.			0
612	Congenital Central Hypoventilation Syndrome (CCHS). , 2021, , 185-195.			0
613	Congenital Central Hypoventilation Syndrome. , 2021, , 239-247.			0
614	Tentative mouse model for the congenital central hypoventilation syndrome: heterozy mutant newborn mice. , 2008, , 243-257.	gous phox2b		1
615	Phox2b and the homeostatic brain. , 2008, , 25-44.			11
616	Congenital central hypoventilation syndrome: from patients to gene discovery. , 2008	, , 45-55.		1
617	Structural and functional brain abnormalities in Congenital Central Hypoventilation Sy 2008, , 57-70.	ndrome. ,		1
618	Polyalanine Tract Disorders and Neurocognitive Phenotypes. Advances in Experimenta Biology, 2012, 769, 185-203.	Medicine and	0.8	18
619	Central Hypoventilation Syndromes. , 2012, , 391-407.			1
620	Neurocristopathies and Particular Associations with Hirschsprungâ \in Ms Disease. , 2008	3, , 253-266.		1
621	The Molecular Genetics of Hirschsprung's Disease. , 2008, , 63-78.			5
622	Genetics of Obstructive Sleep Apnea. , 2005, , 1013-1022.			3
623	Association between <i>PHOX2B</i> gene rs28647582 T>C polymorphism an susceptibility. Bioscience Reports, 2019, 39, .	d Wilms tumor	1.1	4
625	Central hypoventilation with PHOX2B expansion mutation presenting in adulthood. BI 2009, 2009, bcr0920080946-bcr0920080946.	VJ Case Reports,	0.2	2
627	Autonomic neurocristopathy-associated mutations in PHOX2B dysregulate Sox10 exp of Clinical Investigation, 2012, 122, 3145-3158.	ression. Journal	3.9	89

#	Article	IF	CITATIONS
628	NPARM in PHOX2B: why some things just should not be expanded. Journal of Clinical Investigation, 2012, 122, 3056-3058.	3.9	6
629	Case Report: Vocal cord collapse during phrenic nerve-paced respiration in congenital central hypoventilation syndrome. F1000Research, 2012, 1, 42.	0.8	4
630	Hippocampal Volume Reduction in Congenital Central Hypoventilation Syndrome. PLoS ONE, 2009, 4, e6436.	1.1	29
631	Fine Mapping of the NRG1 Hirschsprung's Disease Locus. PLoS ONE, 2011, 6, e16181.	1.1	37
632	The Cerebral Cost of Breathing: An fMRI Case-Study in Congenital Central Hypoventilation Syndrome. PLoS ONE, 2014, 9, e107850.	1.1	26
633	PHOX2B Mutation-Confirmed Congenital Central Hypoventilation Syndrome in a Chinese Family. Chest, 0, , .	0.4	1
634	Pleiotropic effect of common PHOX2B variants in Hirschsprung disease and neuroblastoma. Aging, 2019, 11, 1252-1261.	1.4	12
635	Research progress of neuroblastoma related gene variations. Oncotarget, 2017, 8, 18444-18455.	0.8	29
636	Current Perspectives for the use of Gonane Progesteronergic Drugs in the Treatment of Central Hypoventilation Syndromes. Current Neuropharmacology, 2018, 16, 1433-1454.	1.4	9
637	Molecular Investigations of Sudden Unexplained Deaths. Academic Forensic Pathology, 2011, 1, 194-201.	0.3	7
638	Neurobiology of panic and pH chemosensation in the brain. Dialogues in Clinical Neuroscience, 2011, 13, 475-483.	1.8	54
639	Genetic interactions and modifier genes in Hirschsprung's disease. World Journal of Gastroenterology, 2011, 17, 4937.	1.4	54
640	Depletion of the <i>IKBKAP</i> ortholog in zebrafish leads to hirschsprung disease-like phenotype. World Journal of Gastroenterology, 2015, 21, 2040-2046.	1.4	15
641	Chronic hypoventilation syndromes and sleep-related hypoventilation. Journal of Thoracic Disease, 2015, 7, 1273-85.	0.6	44
642	Significant phenotype variability of congenital central hypoventilation syndrome in a family with polyalanine expansion mutation of the PHOX2B gene. Biomedical Papers of the Medical Faculty of the University Palacký, Olomouc, Czechoslovakia, 2016, 160, 495-498.	0.2	8
643	Late-Onset Hypoventilation Without PHOX2B Mutation or Hypothalamic Abnormalities. Journal of Clinical Sleep Medicine, 2005, 01, 169-172.	1.4	6
644	A Case of "Abnormally Abnormal―Hypoxic Ventilatory Responses: A Novel NPARM <i>PHOX 2B</i> Gene Mutation. Journal of Clinical Sleep Medicine, 2017, 13, 1013-1015.	1.4	6
645	The retrotrapezoid nucleus neurons expressing Atoh1 and Phox2b are essential for the respiratory response to CO2. ELife, 2015, 4, .	2.8	83

		CITATION RE	PORT	
#	Article		IF	CITATIONS
646	Absent phasing of respiratory and locomotor rhythms in running mice. ELife, 2020, 9, .		2.8	17
647	Congenital Central Hypoventilation Syndrome Presenting with Seizures. Cureus, 2020,	12, e6680.	0.2	4
648	Rapid-onset obesity with hypothalamic dysfunction, hypoventilation, and autonomic dy (ROHHAD). , 2021, , .	rsregulation		0
649	Genetic study of a patient with congenital central hypoventilation syndrome in Iran: a c Molecular Biology Reports, 2021, 48, 8239-8243.	case report.	1.0	1
650	Of Mice and Babies: PHOX2B and Obstructive Apneas in Congenital Central Hypoventi American Journal of Respiratory and Critical Care Medicine, 2021, 204, 1128-1130.	lation Syndrome.	2.5	1
651	A Novel Missense Variant of HOXD13 Caused Atypical Synpolydactyly by Impairing the Expression and Literature Review for Genotype–Phenotype Correlations. Frontiers in 12, 731278.	Downstream Gene Genetics, 2021,	1.1	7
652	PEDIATRIC SLEEP AND MEDICAL DISORDERS. , 2004, , 195-208.			0
653	Potential Genetic Contributions to Nonidiopathic, Nonfamilial Pulmonary Hypertension Pulmonary Hypertension, 2005, 4, 24-28.	. Advances in	0.1	0
655	å…^宿€§ä¸æž¢æ€§è,ºèfžä½Žæ•気症候ç¾⊄(ã,ªãf³ãf‡ã,£ãf¼ãfŒã®å'ªã•ॢ) ã®	^y å¨å>½ã,¢ãf³ã,±ãf¼ãf~i	èª ,aedÿ ». Ja∣	oa o ese Journ
656	Types of Sleep-Related Breathing Disorders. , 2008, , 351-363.			0
658	Diaphragmatic Pacing in Children. , 2009, , 523-532.			0
659	Sleep-Disordered Breathing. , 2010, , 1881-1913.			0
660	Sueño, respiración y trastornos neurológicos. , 2011, , 436-498.			0
662	Late Onset Central Hypoventilation Syndrome due to a Heterozygous Polyalanine Repe Mutation in the PHOX2B Gene. Oman Medical Journal, 2011, 26, 356-358.	at Expansion	0.3	1
664	Brain Regulatory Mechanisms Underlying Breathing: Insights for Sleep Pathology. , 201	2, , 461-473.		0
665	Pédiatrie et sommeil. , 2012, , 429-458.			1
666	Molecular genetics of congenital central hypoventilation syndrome and Haddad syndro Genetic Medicine, 2014, 11, 11-15.	me. Journal of	0.1	0
667	Neural Regulation of Lung Development. Pancreatic Islet Biology, 2015, , 43-62.		0.1	0

#	Article	IF	CITATIONS
668	Haddad Syndrome with a Germ-Line Mutation in thePHOX2BGene in a Korean Neonate. Neonatal Medicine, 2015, 22, 162.	0.1	1
671	Genetics and Genomic Basis of Sleep Disorders in Humans. , 2017, , 322-339.e7.		2

672 CLINICAL CASE OF SUCCESSFUL REHABILITATION OF A CHILD WITH UNDINE SYNDROME (CCHS-CONGENITAL) TJ ETOq0 0 0.0gBT /Over

673	Pédiatrie et sommeil. , 2019, , 375-403.		0
674	Congenital Central Hypoventilation Syndrome with Initial Symptom of Pulmonary Hypertension: Case Report and Literature Review. Asian Case Reports in Pediatrics, 2019, 07, 1-6.	0.1	0
676	CONGENITAL CENTRAL HYPOVENTILATION SYNDROME (TWO CASES FROM THE SAME FAMILY). Russian Pediatric Journal, 2019, 20, 180-184.	0.0	2
677	Neuroblastoma Pathogenesis. , 2020, , 29-56.		0
679	Schlafbezogene Atmungsstörungen bei Säglingen und Kindern. , 2020, , 607-616.		0
680	Brainstem Structures Involved in the Generation of Reflex Cough. , 2020, , 45-72.		0
681	Hypothalamic Obesity and Wasting Syndromes. Contemporary Endocrinology, 2021, , 235-280.	0.3	0
682	Genetic Syndromes of Hypothalamic Dysfunction. Contemporary Endocrinology, 2021, , 293-343.	0.3	0
683	Mystery of Expansion: DNA Metabolism and Unstable Repeats. Advances in Experimental Medicine and Biology, 2020, 1241, 101-124.	0.8	3
684	Haddad Syndrome: A Case of Congenital Central Hypoventilation Syndrome Combined with Hirschsprung Disease. Advances in Pediatric Surgery, 2020, 26, 72.	0.2	0
685	Schlafmedizinische Krankheitsbilder im Säglingsalter. , 2020, , 19-32.		0
687	Non-invasive APV in a child with the syndrome of central alveolar hypoventilation. Russian Journal of Pediatric Surgery Anesthesia and Intensive Care, 2020, 9, 78-87.	0.1	2
688	Genes and development of respiratory rhythm generation. , 2008, , 169-189.		0
689	Lessons from mutant newborn mice with respiratory control deficits. , 2008, , 223-241.		0
690	In vitro studies of PHOX2B gene mutations in congenital central hypoventilation syndrome. , 2008, , 71-83.		0

#	Article	IF	CITATIONS
691	Sudden infant death syndrome: study of genes pertinent to cardiorespiratory and autonomic regulation. , 2008, , 85-109.		0
692	Integrating the Genome and Epigenome in Human Disease. , 2009, , 343-368.		0
693	Neuroblastoma and Related Tumors. , 2008, , 233-341.		0
694	Adult cases of late-onset congenital central hypoventilation syndrome and paired-like homeobox 2B-mutation carriers: an additional case report and pooled analysis. Journal of Clinical Sleep Medicine, 2020, 16, 1891-1900.	1.4	10
695	Some congenital diseases may just show up later. Journal of Clinical Sleep Medicine, 2020, 16, 1835-1836.	1.4	0
696	Clinical features, ARIX and PHOX2B nucleotide changes in three families with congenital superior oblique muscle palsy. Acta Medica Okayama, 2008, 62, 45-53.	0.1	4
697	ARIX and PHOX2B polymorphisms in patients with congenital superior oblique muscle palsy. Acta Medica Okayama, 2005, 59, 55-62.	0.1	5
698	Rostral ventrolateral medulla, retropontine region and autonomic regulations. Autonomic Neuroscience: Basic and Clinical, 2022, 237, 102922.	1.4	14
699	Genetic and biological factors in sleep. , 2022, , 73-95.		0
700	Reduced and stable feature sets selection with random forest for neurons segmentation in histological images of macaque brain. Scientific Reports, 2021, 11, 22973.	1.6	2
701	Respiratory disorders of Parkinson's disease. Journal of Neurophysiology, 2022, 127, 1-15.	0.9	11
702	Longâ€read whole genome sequencing reveals HOXD13 alterations in synpolydactyly. Human Mutation, 2022, 43, 189-199.	1.1	7
703	Molecular Mechanisms in Pentanucleotide Repeat Diseases. Cells, 2022, 11, 205.	1.8	14
704	Sleep Disturbances Linked to Genetic Disorders. Sleep Medicine Clinics, 2022, 17, 77-86.	1.2	2
705	Genetic variants in eleven central and peripheral chemoreceptor genes in sudden infant death syndrome. Pediatric Research, 2022, 92, 1026-1033.	1.1	4
707	Trinucleotide CGG Repeat Diseases: An Expanding Field of Polyglycine Proteins?. Frontiers in Genetics, 2022, 13, 843014.	1.1	13
708	Generation of two hiPSC lines (UMILi027-A and UMILi028-A) from early and late-onset Congenital Central hypoventilation Syndrome (CCHS) patients carrying a polyalanine expansion mutation in the PHOX2B gene. Stem Cell Research, 2022, 61, 102781.	0.3	0
709	Advancing genomic technologies and clinical awareness accelerates discovery of disease-associated tandem repeat sequences. Genome Research, 2022, 32, 1-27.	2.4	36

#	Article	IF	CITATIONS
710	The molecular pathogenesis of repeat expansion diseases. Biochemical Society Transactions, 2022, 50, 119-134.	1.6	11
711	Rare cause of neonatal apnea from congenital central hypoventilation syndrome. BMC Pediatrics, 2022, 22, 105.	0.7	1
712	In Transgenic Erythropoietin Deficient Mice, an Increase in Respiratory Response to Hypercapnia Parallels Abnormal Distribution of CO2/H+-Activated Cells in the Medulla Oblongata. Frontiers in Physiology, 2022, 13, 850418.	1.3	4
714	The science of Hirschsprung disease: What we know and where we are headed. Seminars in Pediatric Surgery, 2022, 31, 151157.	0.5	26
716	Schlafbezogene Atmungsstörungen im Kindesalter – Obstruktive Schlafapnoe bei Kindern. , 2022, , 173-176.		0
717	Non-polyalanine repeat mutation in PHOX2B is detected in autopsy cases of sudden unexpected infant death. PLoS ONE, 2022, 17, e0267751.	1.1	1
718	Etonogestrel Administration Reduces the Expression of PHOX2B and Its Target Genes in the Solitary Tract Nucleus. International Journal of Molecular Sciences, 2022, 23, 4816.	1.8	3
719	Perioperative outcomes and the effects of anesthesia in congenital central hypoventilation patients. Sleep and Breathing, 2023, 27, 505-510.	0.9	2
720	Urinary Biomarkers as a Proxy for Congenital Central Hypoventilation Syndrome Patient Follow-Up. Antioxidants, 2022, 11, 929.	2.2	3
721	Advancing therapy for neuroblastoma. Nature Reviews Clinical Oncology, 2022, 19, 515-533.	12.5	97
721 722	Advancing therapy for neuroblastoma. Nature Reviews Clinical Oncology, 2022, 19, 515-533. A Newborn Infant with Congenital Central Hypoventilation Syndrome and Pupillary Abnormalities: A Literature Review. AJP Reports, 0, , .	12.5 0.4	97 0
721 722 724	Advancing therapy for neuroblastoma. Nature Reviews Clinical Oncology, 2022, 19, 515-533. A Newborn Infant with Congenital Central Hypoventilation Syndrome and Pupillary Abnormalities: A Literature Review. AJP Reports, 0, , . Spectrum of pairedâ€like homeobox 2b immunoexpression in pediatric brain tumors with embryonal morphology. Pathology International, 2022, 72, 402-410.	12.5 0.4 0.6	97 0 3
721 722 724 725	Advancing therapy for neuroblastoma. Nature Reviews Clinical Oncology, 2022, 19, 515-533. A Newborn Infant with Congenital Central Hypoventilation Syndrome and Pupillary Abnormalities: A Literature Review. AJP Reports, 0, , . Spectrum of pairedâ€like homeobox 2b immunoexpression in pediatric brain tumors with embryonal morphology. Pathology International, 2022, 72, 402-410. Significance of Serum Oxidative and Antioxidative Status in Congenital Central Hypoventilation Syndrome (CCHS) Patients. Antioxidants, 2022, 11, 1497.	12.5 0.4 0.6 2.2	97 0 3 1
721 722 724 725 726	Advancing therapy for neuroblastoma. Nature Reviews Clinical Oncology, 2022, 19, 515-533. A Newborn Infant with Congenital Central Hypoventilation Syndrome and Pupillary Abnormalities: A Literature Review. AJP Reports, 0, , . Spectrum of pairedâ€like homeobox 2b immunoexpression in pediatric brain tumors with embryonal morphology. Pathology International, 2022, 72, 402-410. Significance of Serum Oxidative and Antioxidative Status in Congenital Central Hypoventilation syndrome (CCHS) Patients. Antioxidants, 2022, 11, 1497. Molecular Organization and Patterning of the Medulla Oblongata in Health and Disease. International Journal of Molecular Sciences, 2022, 23, 9260.	12.5 0.4 0.6 2.2 1.8	97 0 3 1 8
721 722 724 725 726 727	Advancing therapy for neuroblastoma. Nature Reviews Clinical Oncology, 2022, 19, 515-533. A Newborn Infant with Congenital Central Hypoventilation Syndrome and Pupillary Abnormalities: A Literature Review. AJP Reports, 0, , . Spectrum of pairedâ€like homeobox 2b immunoexpression in pediatric brain tumors with embryonal morphology. Pathology International, 2022, 72, 402-410. Significance of Serum Oxidative and Antioxidative Status in Congenital Central Hypoventilation Syndrome (CCHS) Patients. Antioxidants, 2022, 11, 1497. Molecular Organization and Patterning of the Medulla Oblongata in Health and Disease. International Journal of Molecular Sciences, 2022, 23, 9260. Super-enhancer-associated TTC8 alters the nucleocytoplasmic distribution of PHOX2B and activates MAPK signaling in neuroblastoma. Genes and Diseases, 2022,	12.5 0.4 0.6 2.2 1.8 1.5	97 0 3 1 8 1
 721 722 724 725 726 727 728 	Advancing therapy for neuroblastoma. Nature Reviews Clinical Oncology, 2022, 19, 515-533. A Newborn Infant with Congenital Central Hypoventilation Syndrome and Pupillary Abnormalities: A Literature Review. AJP Reports, 0, , . Spectrum of pairedâ€like homeobox 2b immunoexpression in pediatric brain tumors with embryonal morphology. Pathology International, 2022, 72, 402-410. Significance of Serum Oxidative and Antioxidative Status in Congenital Central Hypoventilation Syndrome (CCHS) Patients. Antioxidants, 2022, 11, 1497. Molecular Organization and Patterning of the Medulla Oblongata in Health and Disease. International Journal of Molecular Sciences, 2022, 23, 9260. Super-enhancer-associated TTC8 alters the nucleocytoplasmic distribution of PHOX2B and activates MAPK signaling in neuroblastoma. Genes and Diseases, 2022, Early development of the breathing network. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2022, .125-149.	12.5 0.4 0.6 2.2 1.8 1.5 1.0	97 0 3 1 8 1 1
 721 722 724 725 726 727 728 729 	Advancing therapy for neuroblastoma. Nature Reviews Clinical Oncology, 2022, 19, 515-533. A Newborn Infant with Congenital Central Hypoventilation Syndrome and Pupillary Abnormalities: A Literature Review. AJP Reports, 0, , . Spectrum of pairedà&kike homeobox 2b immunoexpression in pediatric brain tumors with embryonal morphology. Pathology International, 2022, 72, 402-410. Significance of Serum Oxidative and Antioxidative Status in Congenital Central Hypoventilation Syndrome (CCHS) Patients. Antioxidants, 2022, 11, 1497. Molecular Organization and Patterning of the Medulla Oblongata in Health and Disease. International Journal of Molecular Sciences, 2022, 23, 9260. Super-enhancer-associated TTC8 alters the nucleocytoplasmic distribution of PHOX2B and activates MAPK signaling in neuroblastoma. Genes and Diseases, 2022, . Early development of the breathing network. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2022, 125-149. Congenital central alveolar hypoventilation syndrome. , 2022,	12.5 0.4 0.6 2.2 1.8 1.5 1.0	97 0 3 1 8 1 4 0

ARTICLE IF CITATIONS # Developmental disorders affecting the respiratory system: CCHS and ROHHAD. Handbook of Clinical 731 1.0 9 Neurology / Edited By P J Vinken and G W Bruyn, 2022, , 53-91. Embryology and anatomy of Hirschsprung disease. Seminars in Pediatric Surgery, 2022, 31, 151227. Phox2b mutation mediated by Atoh1 expression impaired respiratory rhythm and ventilatory responses 733 2.8 6 to hypoxia and hypercapnia. ÉLife, 0, 11, . Transitional care and clinical management of adolescents, young adults, and suspected new adult 734 patients with congenital central hypoventilation syndrome. Clinical Autonomic Research, 2023, 33, 231-249. Transcription factors regulating the specification of brainstem respiratory neurons. Frontiers in 736 1.4 6 Molecular Neuroscience, 0, 15, . Comparative physiological anthropogeny: exploring molecular underpinnings of distinctly human phenotypes. Physiological Reviews, 2023, 103, 2171-2229. 13.1 738 Congenital central hypoventilation syndrome (CCHS) and PHOX2B pathogenic variants., 2023, 515-520. 0 Genetics of Hirschsprung's disease. Pediatric Surgery International, 2023, 39, . Case Report: A novel PHOX2B p.Ala248 Ala266dup variant causing congenital central hypoventilation 740 0.9 1 syndrome. Frontiers in Pediatrics, 0, 10, Serotonin and the ventilatory effects of etonogestrel, a gonane progestin, in a murine model of 741 1.5 congenital central hypoventilation syndrome. Frontiers in Endocrinology, 0, 14, . 745 Chronic Neurological Disorders., 2023, , 227-245. 0 Chronic Noninvasive Ventilatory Support in Various Chronic Respiratory Conditions Including 746 Protocols., 2023,, 131-144. Rare diseases of respiratory drive., 2023, , 357-366. 747 0 Rapid-onset obesity with hypothalamic dysfunction, hypoventilation, and autonomic dysregulation (ROHHAD): a collaborative review of the current understanding. Clinical Autonomic Research, 2023, 1.4 33, 251-268. Development and Developmental Disorders of the Brain Stem., 2023, , 445-521. 756 0 Non-Invasive Ventilation in Pediatric Patients., 2023, , 1-19. 759 Congenital Central Hypoventilation Syndrome., 2023, , . 0 766